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Supplementary Table 1

Identifier	Gene	RefSeq transcript	cDNA position	Amino acid position	Chromosomal position	Zygosity	Inheritance pattern	Genoox classification	ACMG evidence criteria	HGMD ID	ClinVar ID	gnomAD AF	Transmission/Phase	Prescreening	Family history	Gender	Average depth
Solved cases																	
ACHM 1213	***CNGA3***	**NM_001298.2**	**c.489_492del**	**p.(Ser164Alafs*8)**	**NC_000002.11:g.99006160_99006163del**	**het**	**AR**	**P**	**PM2, PVS1, PP5**	-	-	-	**unknown**	**no**	**unknown**	**f**	**47.82**
ACHM 1213	***CNGA3***	**NM_001298.2**	**c.778dup**	**p.(Asp260Glyfs*27)**	**NC_000002.11:g.99012411dup**	**het**	**AR**	**P**	**PM2, PVS1_STRONG, PP5_STRONG**	-	-	-	**unknown**	**no**	**unknown**	**f**	**47.82**
ACHM 1220	***CNGB3***	**NM_019098.4**	**c.1148del**	**p.(Thr383Ilefs*13)**	**NC_000008.10:g.87656009del**	**het**	**AR**	**P**	**PM2, PVS1, PP5, PS4_MODERATE**	**CD001927**	**5225**	**0.0020**	**unknown**	**no**	**negative**	**f**	**55.86**
ACHM 1220	***CNGB3***	**NM_019098.4**	**c.1578+1G>A**	**p.(?)**	**NC_000008.10:g.87638210C>T**	**het**	**AR**	**P**	**PM2, PVS1, PP5**	**CS002123**	**189031**	**0.0001**	**unknown**	**no**	**negative**	**f**	**55.86**
ACHM 1229	***CNGB3***	**NM_019098.4**	**c.1148del**	**p.(Thr383Ilefs*13)**	**NC_000008.10:g.87656009del**	**het**	**AR**	**P**	**PM2, PVS1, PP5, PS4_MODERATE**	**CD001927**	**5225**	**0.0020**	**unknown**	**no**	**negative**	**f**	**62.60**
ACHM 1229	***CNGB3***	**NM_019098.4**	**c.852+4013_903+1698dup**	**p.(?)**	**NC_000008.10:g.87664544_87675142dup**	**het**	**AR**	**LP**	**PM2, PVS1**	**na**	-	**na**	**unknown**	**no**	**negative**	**f**	**62.60**
ACHM 1261	***CNGB3***	**NM_019098.4**	**c.1148del**	**p.(Thr383Ilefs*13)**	**NC_000008.10:g.87656009del**	**hom**	**AR**	**P**	**PM2, PVS1, PP5, PS4_MODERATE**	**CD001927**	**5225**	**0.0020**	**unknown**	**no**	**unknown**	**f**	**55.36**
ACHM 1271	***CNGA3***	**NM_001298.2**	**c.830G>A**	**p.(Arg277His)**	**NC_000002.11:g.99012463G>A**	**het**	**AR**	**P**	**PS1_MODERATE, PP3, PM2, PM5, PM1, PP2, PPS_STRONG**	**CM014539**	-	**0.0001**	**unknown**	**no**	**positive (duplex)**	**f**	**28.30**
ACHM 1271	***CNGA3***	**NM_001298.2**	**c.985G>T**	**p.(Gly329Cys)**	**NC_000002.11:g.99012618G>T**	**het**	**AR**	**P**	**PP3, PM2, PM1, PP2, PPS_STRONG**	**CM1110706**	-	-	**unknown**	**no**	**positive (duplex)**	**f**	**28.30**
ACHM 1272	***CNGA3***	**NM_001298.2**	**c.822G>T**	**p.(Arg274Ser)**	**NC_000002.11:g.99012455G>T**	**het**	**AR**	**LP**	**PP3, PM2, PM1, PP2**	**CM102644**	-	**0.0001**	**unknown**	**no**	**negative**	**f**	**53.94**
ACHM 1272	***CNGB3***	**NM_019098.4**	**c.1208G>A**	**p.(Arg403Gln)**	**NC_000008.10:g.87645092C>T**	**hom**	**AR**	**VUS**	**PP3, PP5, BS1, BS2, BP6**	**CM043918**	-	**0.0046**	**unknown**	**no**	**negative**	**f**	**53.94**
ACHM 1280	***CNGB3***	**NM_019098.4**	**c.1148del**	**p.(Thr383Ilefs*13)**	**NC_000008.10:g.87656009del**	**het**	**AR**	**P**	**PM2, PVS1, PP5, PS4_MODERATE**	**CD001927**	**5225**	**0.0020**	**unknown**	**single genes**	**unknown**	**f**	**45.68**
ACHM 1280	***CNGB3***	**NM_019098.4**	**c.1208G>A**	**p.(Arg403Gln)**	**NC_000008.10:g.87645092C>T**	**het**	**AR**	**VUS**	**PP3, PP5, BS1, BS2, BP6**	**CM043918**	-	**0.0046**	**unknown**	**single genes**	**unknown**	**f**	**45.68**
ACHM 1281	***CNGB3***	**NM_019098.4**	**c.1148del**	**p.(Thr383Ilefs*13)**	**NC_000008.10:g.87656009del**	**hom**	**AR**	**P**	**PM2, PVS1, PP5, PS4_MODERATE**	**CD001927**	**5225**	**0.0020**	**unknown**	**no**	**negative**	**f**	**50.85**
ACHM 1281	*CNGA3*	NM_001298.2	c.829C>T	p.(Arg277Cys)	NC_000002.11:g.99012462C>T	het	AR	P	PP3, PM2, PM5, PM1, PP2, PP5_STRONG	CM014538	-	0.0001	unknown	no	negative	f	50.85
ACHM 1282	***CNGB3***	**NM_019098.4**	**c.1148del**	**p.(Thr383Ilefs*13)**	**NC_000008.10:g.87656009del**	**hom**	**AR**	**P**	**PM2, PVS1, PP5, PS4_MODERATE**	**CD001927**	**5225**	**0.0020**	**paternal/maternal**	**no**	**negative**	**m**	**49.11**
ACHM 1287	***CNGB3***	**NM_019098.4**	**c.1578+1G>A**	**p.(?)**	**NC_000008.10:g.87638210C>T**	**het**	**AR**	**P**	**PM2, PVS1, PP5**	**CS002123**	-	**0.0001**	**unknown**	**no**	**negative**	**f**	**50.59**
ACHM 1287	***CNGB3***	**NM_019098.4**	**c.1208G>A**	**p.(Arg403Gln)**	**NC_000008.10:g.87645092C>T**	**het**	**AR**	**VUS**	**PP3, PP5, BS1, BS2, BP6**	**CM043918**	-	**0.0046**	**unknown**	**no**	**negative**	**f**	**50.59**
ACHM 1290	***CNGB3***	**NM_019098.4**	**c.1148del**	**p.(Thr383Ilefs*13)**	**NC_000008.10:g.87656009del**	**het**	**AR**	**P**	**PM2, PVS1, PP5, PS4_MODERATE**	**CD001927**	**5225**	**0.0020**	**paternal**	**no**	**negative**	**m**	**56.20**
ACHM 1290	***CNGB3***	**NM_019098.4**	**c.1179-2A>T**	**p.(?)**	**NC_000008.10:g.87645123T>A**	**het**	**AR**	**P**	**PM2, PVS1, PP5**	-	**371304**	-	**maternal**	**no**	**negative**	**m**	**56.20**
ACHM 1291	***CNGA3***	**NM_001298.2**	**c.1641C>A**	**p.(Phe547Leu)**	**NC_000002.11:g.99013274C>A**	**het**	**AR**	**P**	**PS1_MODERATE, PP3, PM2, PM5_SUPPORTING, PM1, PP2, PPS_STRONG**	**CM980380**	**9478**	**0.0002**	**unknown**	**no**	**negative**	**m**	**55.08**
ACHM 1291	***CNGA3***	**NM_001298.2**	**c.827A>G**	**p.(Asn276Ser)**	**NC_000002.11:g.99012460A>G**	**het**	**AR**	**P**	**PP3, PM2, PM1, PP2, PPS_STRONG**	**CM118229**	**156335**	-	**unknown**	**no**	**negative**	**m**	**55.08**
ACHM 1291	*ABCA4*	NM_000350.2	c.5882G>A	p.(Gly1961Glu)	NC_000001.10:g.94473807C>T	het	AR	P	PP3, PM2, PM5, PP2, PPS_STRONG, PS4_MODERATE	CM970016	635154	0.0047	unknown	no	negative	m	55.08
ACHM 1297	***CNGA3***	**NM_001298.2**	**c.1641C>A**	**p.(Phe547Leu)**	**NC_000002.11:g.99013274C>A**	**het**	**AR**	**P**	**PS1_MODERATE, PP3, PM2, PM5_SUPPORTING, PM1, PP2, PPS_STRONG**	**CM980380**	-	**0.00016**	**unknown**	**no**	**negative**	**f**	**37.58**
ACHM 1297	***CNGA3***	**NM_001298.2**	**c.1360A>T**	**p.(Lys454*)**	**NC_000002.11:g.99012993A>T**	**het**	**AR**	**P**	**PM2, PVS1, PP5**	-	-	-	**unknown**	**no**	**negative**	**f**	**37.58**
ACHM 1297	*ABCA4*	NM_000350.2	c.5146G>A	p.(Gly1716Ser)	NC_000001.10:g.94568627C>T	het	AR	VUS	PM2	CM032798	-	0.0004	unknown	no	negative	f	37.58
ACHM 1297	*ABCA4*	NM_000350.2	c.676C>A	p.(Arg226Ser)	NC_000001.10:g.94564442G>T	het	AR	VUS	PM2	CM213078	-	0.00007	unknown	no	negative	f	37.58
ACHM 1297	*ABCA4*	NM_000350.2	c.5603A>T	p.(Asn1868Ile)	NC_000001.10:g.94476467T>A	het	AR	VUS	PP2, PP5, BA1, BS2	CM015091	-	0.04	unknown	no	negative	f	37.58
ACHM 1298	***CNGA3***	**NM_001298.2**	**c.1641C>A**	**p.(Phe547Leu)**	**NC_000002.11:g.99013274C>A**	**het**	**AR**	**P**	**PS1_MODERATE, PP3, PM2, PM5_SUPPORTING, PM1, PP2, PPS_STRONG**	**CM980380**	**9478**	**0.0002**	**unknown**	**no**	**negative**	**m**	**37.81**
ACHM 1298	***CNGA3***	**NM_001298.2**	**c.778G>A**	**p.(Asp260Asn)**	**NC_000002.11:g.99012411G>A**	**het**	**AR**	**LP**	**PP3, PM2, PP2, PPS_STRONG**	**CM014536**	**689726**	**0.0001**	**unknown**	**no**	**negative**	**m**	**37.81**
ACHM 1300	***CNGA3***	**NM_001298.2**	**c.1749del**	**p.(Leu584Serfs*23)**	**NC_000002.11:g.99013382del**	**het**	**AR**	**P**	**PM2, PVS1, PP5, PS4_MODERATE**	-	-	-	**unknown**	**no**	**negative**	**f**	**40.18**
ACHM 1300	***CNGA3***	**NM_001298.2**	**c.848G>A**	**p.(Arg283Gln)**	**NC_000002.11:g.99012481G>A**	**het**	**AR**	**P**	**PS1_MODERATE, PP3, PM2, PM5, PM1, PP2, PPS_STRONG**	**CM980375**	-	**0.00006**	**unknown**	**no**	**negative**	**f**	**40.18**
ACHM 1301	***CNGA3***	**NM_001298.2**	**c.1021T>C**	**p.(Ser341Pro)**	**NC_000002.11:g.99012654T>C**	**het**	**AR**	**P**	**PP3, PM2, PM1, PP2, PPS_STRONG**	**CM014541**	**866559**	**0.0001**	**unknown**	**no**	**negative**	**m**	**40.82**
ACHM 1301	***CNGA3***	**NM_001298.2**	**c.847C>T**	**p.(Arg283Trp)**	**NC_000002.11:g.99012480C>T**	**het**	**AR**	**P**	**PS1_MODERATE, PP3, PM2, PM5, PM1, PP2, PPS_STRONG**	**CM980376**	**9474**	**0.0001**	**unknown**	**no**	**negative**	**m**	**40.82**
ACHM 1302	***CNGB3***	**NM_019098.4**	**c.1148del**	**p.(Thr383Ilefs*13)**	**NC_000008.10:g.87656009del**	**hom**	**AR**	**P**	**PM2, PVS1, PP5, PS4_MODERATE**	**CD001927**	**5225**	**0.0020**	**unknown**	**no**	**negative**	**f**	**41.63**
ADRP 15	***SNRNP200***	**NM_014014.4**	**c.2041C>T**	**p.(Arg681Cys)**	**NC_000002.11:g.96958829G>A**	**het**	**AD**	**P**	**PP3, PM2, PM5, PM1_SUPPORTING, PP2, PPS_STRONG**	**CM113957**	-	**0.0001**	**unknown**	**single genes**	**positive (multiplex)**	**f**	**54.36**
ADRP 15	*RP11*	NM_178857.5	c.6299G>C	p.(Gly2100Ala)	NC_000008.10:g.10465309C>G	het	AD+AR	VUS	PM2, BP4	-	-	-	unknown	single genes	positive (multiplex)	f	54.36
ADRP 15	*RP11*	NM_178857.5	c.945G>A	p.(Met315Ile)	NC_000008.10:g.10470663C>T	het	AD+AR	VUS	PM2, BP4	-	-	0.0001	unknown	single genes	positive (multiplex)	f	54.36
ADRP 18	***NR2E3***	**NM_014249.3**	**c.166G>A**	**p.(Gly56Arg)**	**NC_000015.9:g.72103870G>A**	**het**	**AR+AD**	**P**	**PS1, PM2, PM1_SUPPORTING, PPS_STRONG**	**CM073246**	-	-	**unknown**	**Panel**	**positive (multiplex)**	**f**	**55.01**
ADRP 18	*CEP290*	NM_025114.3	c.1709C>G	p.(Ser570*)	NC_000012.11:g.88512262G>C	het	AR	P	PM2, PVS1, PP5	CM071592	-	0.0001	unknown	Panel	positive (multiplex)	f	55.01
ADRP 18	*IFT140*	NM_014714.3	c.454C>T	p.(Leu152Phe)	NC_000016.9:g.1642505G>A	het	AR	VUS	PM2, PP5	CM136696	-	0.0001	unknown	Panel	positive (multiplex)	f	55.01
ADRP 115	***PRPF31***	**NM_015629.3**	**c.239-2A>G**	**p.(?)**	**NC_000019.9:g.54625237A>G**	**het**	**AD**	**P**	**PM2, PVS1_STRONG, PPS_STRONG**	**CS2020272**	-	-	**unknown**	**single genes**	**positive (duplex)**	**f**	**57.80**
ADRP 221-1	***PRPF8***	**NM_006445.3**	**c.6910T>C**	**p.(Phe2304Leu)**	**NC_000017.10:g.1554194A>G**	**het**	**AD**	**P**	**PS1, PP3, PM2, PM1, PP2, PPS**	**CM149179**	-	-	**unknown**	**Panel**	**positive (multiplex)**	**f**	**77.72**
ADRP 221-1	*PRPF6*	NM_012469.3	c.2200C>G	p.(Gln734Glu)	NC_000020.10:g.62658506C>G	het	AD	VUS	PM2	-	-	0.0001	unknown	Panel	positive (multiplex)	f	77.72
ADRP 221-2	***PRPF8***	**NM_006445.3**	**c.6910T>C**	**p.(Phe2304Leu)**	**NC_000017.10:g.1554194A>G**	**het**	**AD**	**P**	**PS1, PP3, PM2, PM1, PP2, PPS**	**CM149179**	-	-	**unknown**	**Panel**	**positive (multiplex)**	**m**	**49.41**
ADRP 221-2	*PRPF6*	NM_012469.3	c.2200C>G	p.(Gln734Glu)	NC_000020.10:g.62658506C>G	het	AD	VUS	PM2	-	-	0.0001	unknown	Panel	positive (multiplex)	m	49.41
ADRP 287	***CRX***	**NM_000554.4**	**na**	**na**	**NC_000019.9:g.48309247_48350581del**	**het**	**AD**	**LP**	**PM2, PVS1**	**na**	-	**na**	**maternal**	**no**	**positive (duplex)**	**f**	**45.30**
ADRP 287	*USH2A*	NM_206933.2	c.7121-1G>T	p.(?)	NC_000001.10:g.216108138C>A	het	AR	LP	PM2, PVS1_STRONG	-	-	-	unknown	no	positive (duplex)	f	45.30
ADRP 353	***PRPF31***	**NM_015629.3**	**na**	**na**	**NC_000019.9:g.54627731_54636213del**	**het**	**AD**	**LP**	**PM2, PVS1**	**na**	-	**na**	**unknown**	**no**	**positive (triplex)**	**f**	**56.34**
ADRP 367	***PRPF31***	**NM_015629.3**	**c.358_359del**	**p.(Lys120Glufs*4)**	**NC_000019.9:g.54625911_54625912del**	**het**	**AD**	**P**	**PM2, PVS1, PP5**	**HD060013**	**1275773**	-	**unknown**	**Panel**	**positive (multiplex)**	**f**	**38.53**
ADRP 367	*NR2E3*	NM_014249.3	c.227G>A	p.(Arg76Gln)	NC_000015.9:g.72103931G>A	het	AR+AD	LP	PM2, PM5, PM1_SUPPORTING, PP5	CM000531	-	0.0003	unknown	Panel	positive (multiplex)	f	38.53
ADRP 386-1	***C1QTNF5***	**NM_001278431.1**	**c.538C>G**	**p.(Gln180Glu)**	**NC_000011.9:g.119210235G>C**	**het**	**AD**	**P**	**PM2, PS3, PP1_STRONG**	**CM217871**	-	-	**unknown**	**Panel**	**positive (multiplex)**	**f**	**45.38**
ADRP 386-2	***C1QTNF5***	**NM_001278431.1**	**c.538C>G**	**p.(Gln180Glu)**	**NC_000011.9:g.119210235G>C**	**het**	**AD**	**P**	**PM2, PS3, PP1_STRONG**	**CM217871**	-	-	**unknown**	**Panel**	**positive (multiplex)**	**m**	**45.22**
ADRP 457	***IMPDH1***	**NM_000883.3**	**c.931G>A**	**p.(Asp311Asn)**	**NC_000007.13:g.128038611C>T**	**het**	**AD**	**P**	**PP3, PM2, PM1_SUPPORTING, PPS_STRONG**	**CM020283**	-	**0.0001**	**unknown**	**no**	**positive (duplex)**	**f**	**46.41**
ADRP 467	***RP1***	**NM_006269.1**	**c.2332A>T**	**p.(Lys778*)**	**NC_000008.10:g.55538774A>T**	**het**	**AD+AR**	**P**	**PM2, PVS1_STRONG, PPS_STRONG**	**CM022060**	**845845**	-	**unknown**	**no**	**positive (multiplex)**	**f**	**41.37**
ADRP 467	*FBLN5*	NM_006329.3	c.799G>A	p.(Gly267Ser)	NC_000014.8:g.92349361C>T	het	AD	VUS	PP3, PM2	CM062699	-	0.00027	unknown	no	positive (multiplex)	f	41.37
ADRP 470-1	***RP1***	**NM_006269.1**	**c.2161del**	**p.(Gly723Glufs*15)**	**NC_000008.10:g.55538603del**	**het**	**AD+AR**	**P**	**PM2, PVS1_STRONG, PPS_STRONG**	**CD160487**	**810288**	**0.000004**	**unknown**	**no**	**positive (multiplex)**	**m**	**64.72**
ADRP 470-2	***RP1***	**NM_006269.1**	**c.2161del**	**p.(Gly723Glufs*15)**	**NC_000008.10:g.55538603del**	**het**	**AD+AR**	**P**	**PM2, PVS1_STRONG, PPS_STRONG**	**CD160487**	**810288**	**0.000004**	**unknown**	**no**	**positive (multiplex)**	**m**	**48.44**
ADRP 473	***PRPF31***	**NM_015629.3**	**c.217A>T**	**p.(Lys73*)**	**NC_000019.9:g.54621992A>T**	**het**	**AD**	**P**	**PM2, PVS1, PP5**	**CM1312814**	-	-	**maternal**	**no**	**positive (multiplex)**	**m**	**48.38**
ADRP 473	*ABCA4*	NM_000350.2	c.1928T>G	p.(Val643Gly)	NC_000001.10:g.94528142A>C	het	AR	VUS	PP3, PM5_SUPPORTING, PP2, PP5	CM074661	-	0.0017	unknown	no	positive (multiplex)	m	48.38
ADRP 475	***PRPH2***	**NM_000322.4**	**c.634A>G**	**p.(Ser212Gly)**	**NC_000006.11:g.42672297T>C**	**het**	**AD**	**P**	**PP3, PM2, PM5, PM1_STRONG, PP2, PP5, PS4_MODERATE**	**CM920606**	-	-	**unknown**	**no**	**positive (triplex)**	**f**	**46.81**
ADRP 478	***PRPF31***	**NM_015629.3**	**c.238+515_1375-917del**	**p.(?)**	**NC_000019.9:g.54622528_54633821del**	**het**	**AD**	**P**	**PM2, PVS1, PP5**	**na**	-	**na**	**unknown**	**no**	**positive (duplex)**	**m**	**49.95**
ADRP 482	***PRPF31***	**NM_015629.3**	**c.946-2A>C**	**p.(?)**	**NC_000019.9:g.54631446A>C**	**het**	**AD**	**P**	**PM2, PVS1, PP5**	-	-	-	**unknown**	**no**	**positive (multiplex)**	**m**	**51.87**
ADRP 483	***PRPF31***	**NM_015629.3**	**c.238+515_1375-917del**	**p.(?)**	**NC_000019.9:g.54622528_54633821del**	**het**	**AD**	**P**	**PM2, PVS1, PP5**	**na**	-	**na**	**unknown**	**no**	**positive (multiplex)**	**m**	**44.21**
ADRP 485	***PRPF31***	**NM_015629.3**	**na**	**na**	**NC_000019.9:g.54615301_54637092del**	**het**	**AD**	**P**	**PM2, PVS1, PP5**	**na**	-	**na**	**unknown**	**no**	**positive (duplex)**	**m**	**50.02**
ADRP 488	***BEST1***	**NM_004183.3**	**c.713A>G**	**p.(Gln238Arg)**	**NC_000011.9:g.61724935A>G**	**het**	**AD+AR**	**P**	**PP3, PM2, PM1, PP2, PPS_STRONG**	-	-	-	**unknown**	**single genes**	**positive (duplex)**	**m**	**49.43**
ADRP 490	***PRPF31***	**NM_015629.3**	**na**	**na**	**NC_000019.9:g.54617756_54622314del**	**het**	**AD**	**P**	**PM2, PVS1, PP5**	**na**	-	**na**	**unknown**	**no**	**positive (duplex)**	**f**	**55.49**
ADRP 491	***RHO***	**NM_000539.3**	**c.539C>G**	**p.(Pro180Arg)**	**NC_000003.11:g.129251102C>G**	**het**	**AR+AD**	**LP**	**PM2, PM5, PM1_STRONG, PP2**	-	-	-	**unknown**	**no**	**positive (multiplex)**	**m**	**50.96**
ADRP 495	***RHO***	**NM_000539.3**	**c.1040C>T**	**p.(Pro347Leu)**	**NC_000003.11:g.129252554C>T**	**het**	**AR+AD**	**P**	**PM2, PM5, PM1_STRONG, PP2, PP5, PS4_MODERATE**	**CM900199**	**13014**	**0.0001**	**unknown**	**no**	**positive (multiplex)**	**f**	**49.58**
ADRP 496	***PRPF8***	**NM_006445.3**	**c.6970dup**	**p.(Glu2324Glyfs*61)**	**NC_000017.10:g.1554138dup**	**het**	**AD**	**P**	**PM2, PVS1_STRONG, PPS_STRONG**	**CI2036755**	-	-	**unknown**	**single genes**	**positive (multiplex)**	**f**	**48.07**
ADRP 496	*USH2A*	NM_206933.2	c.14912G>A	p.(Arg4971Gln)	NC_000001.10:g.215813956C>T	het	AR	VUS	PM2, BP4	-	-	0.0001	unknown	single genes	positive (multiplex)	f	48.07
ADRP 496	*USH2A*	NM_206933.2	c.2276G>T	p.(Cys759Phe)	NC_000001.10:g.216420460C>A	het	AR	P	PP3, PM2, PM1_SUPPORTING, PP5_STRONG, PS4_MODERATE	CM001372	2356	0.0010	unknown	single genes	positive (multiplex)	f	48.07
ADRP 497	***RHO***	**NM_000539.3**	**c.50C>T**	**p.(Thr17Met)**	**NC_000003.11:g.129247626C>T**	**het**	**AR+AD**	**P**	**PM2, PM5, PM1, PP2, PPS_STRONG**	**CM910325**	**13018**	**0.0001**	**unknown**	**no**	**positive (duplex)**	**f**	**55.03**
ADRP 499	***RHO***	**NM_000539.3**	**c.512C>G**	**p.(Pro171Arg)**	**NC_000003.11:g.129249869C>G**	**het**	**AR+AD**	**LP**	**PS1_MODERATE, PM2, PM5, PM1, PP2**	**CM1723362**	-	-	**unknown**	**no**	**positive (triplex)**	**m**	**46.82**
ADRP 499	*USH2A*	NM_206933.2	c.14426C>G	p.(Thr4809Ser)	NC_000001.10:g.215822026G>C	het	AR	VUS	PM2, PM5	-	-	-	unknown	no	positive (triplex)	m	46.82
ADRP 499	*USH2A*	NM_206933.2	c.1651C>T	p.(Arg551Cys)	NC_000001.10:g.216465706G>A	het	AR	VUS	PM2	-	-	0.0001	unknown	no	positive (triplex)	m	46.82
ADRP 500	***RHO***	**NM_000539.3**	**c.1032G>C**	**p.(Gln344His)**	**NC_000003.11:g.129252546G>C**	**het**	**AR+AD**	**P**	**PM2, PM1_STRONG, PP2, PPS_STRONG**	**CM211897**	-	**0.0001**	**unknown**	**no**	**positive (triplex)**	**m**	**52.93**
ADRP 501	***PRPH2***	**NM_000322.4**	**c.646C>T**	**p.(Pro216Ser)**	**NC_000006.11:g.42672285G>A**	**het**	**AD**	**P**	**PP3, PM2, PM5, PM1_STRONG, PP2, PP5**	**CM941212**	-	-	**unknown**	**no**	**positive (duplex)**	**m**	**45.27**
ADRP 504	***TOPORS***	**NM_005802.4**	**c.2558_2559insT**	**p.(His855Thrfs*18)**	**NC_000009.11:g.32541964_32541965insA**	**het**	**AD**	**P**	**PM2, PVS1_STRONG, PPS_STRONG**	-	-	-	**unknown**	**no**	**positive (triplex)**	**m**	**44.93**
ADRP 505	***PRPF31***	**NM_015629.3**	**c.395del**	**p.(Ala132Aspfs*66)**	**NC_000019.9:g.54625948del**	**het**	**AD**	**P**	**PM2, PVS1, PP5**	-	-	-	**unknown**	**no**	**unknown**	**f**	**60.64**
ADRP 505	*ABCA4*	NM_000350.2	c.2588G>C	p.(Gly863Ala)	NC_000001.10:g.94517254C>G	het	AR	P	PP3, PM2, PP2, PPS_STRONG, PS4_MODERATE	CM970003&CS024003	-	0.0044	unknown	no	unknown	f	60.64
ADRP 508	***PRPF31***	**NM_015629.3**	**c.1126_1129dup**	**p.(Arg377Glnfs*99)**	**NC_000019.9:g.54631732_54631735dup**	**het**	**AD**	**P**	**PM2, PVS1, PP5**	-	**1184921**	-	**paternal**	**no**	**positive (multiplex)**	**f**	**56.89**
ADRP 513	***C1QTNF5***	**NM_001278431.1**	**c.489C>G**	**p.(Ser163Arg)**	**NC_000011.9:g.119210284G>C**	**het**	**AD**	**P**	**PS1, PM2, PP3_STRONG**	**CM032554**	**2126**	-	**unknown**	**no**	**positive (multiplex)**	**m**	**59.08**
ADRP 515	***RHO***	**NM_000539.3**	**c.316G>A**	**p.(Gly106Arg)**	**NC_000003.11:g.129247892G>A**	**het**	**AD+AR**	**P**	**PS1, PM2, PM5, PM1, PP2, PP5**	**CM920609**	**13038**	**0.0001**	**unknown**	**no**	**positive (duplex)**	**m**	**58.33**
ADRP 515	*PROM1*	NM_006017.2	c.2490-2A>G	p.(?)	NC_000004.11:g.15981529T>C	het	AD+AR	P	PM2, PVS1_STRONG, PPS_STRONG	-	-	0.0001	unknown	no	positive (duplex)	m	58.33
ADRP 516	***PRPF31***	**NM_015629.3**	**c.1462_1472del**	**p.(Lys488Argfs*75)**	**NC_000019.9:g.54634825_54634835del**	**het**	**AD**	**LP**	**PM2, PVS1_MODERATE, PPS_STRONG**	**CD188477**	**1275813**	-	**unknown**	**no**	**unknown**	**m**	**42.22**
ADRP 516	*PCDH15*	NM_033056.3	c.366del	p.(Asn122Lysfs*14)	NC_000010.10:g.56128988del	het	AR	P	PM2, PVS1, PP5	-	-	-	unknown	no	unknown	m	42.22
ADRP 518-1	***KLHL7***	**NM_001031710.2**	**c.458C>T**	**p.(Ala153Val)**	**NC_000007.13:g.23180403C>T**	**het**	**AD**	**P**	**PP3, PM2, PM5_SUPPORTING, PM1, PP2, PPS_STRONG**	**CM093480**	**1009**	-	**unknown**	**single genes**	**positive (duplex)**	**f**	**55.62**
ADRP 518-2	***KLHL7***	**NM_001031710.2**	**c.458C>T**	**p.(Ala153Val)**	**NC_000007.13:g.23180403C>T**	**het**	**AD**	**P**	**PP3, PM2, PM5_SUPPORTING, PM1, PP2, PPS_STRONG**	**CM093480**	**1009**	-	**unknown**	**single genes**	**positive (duplex)**	**m**	**36.40**
ADRP 520	***RP1***	**NM_006269.1**	**c.2332A>T**	**p.(Lys778*)**	**NC_000008.10:g.55538774A>T**	**het**	**AD+AR**	**P**	**PM2, PVS1_STRONG, PPS_STRONG**	**CM022060**	-	-	**unknown**	**Panel**	**positive (duplex)**	**m**	**37.14**
ADRP 521	***RHO***	**NM_000539.3**	**c.1040C>T**	**p.(Pro347Leu)**	**NC_000003.11:g.129252554C>T**	**het**	**AR+AD**	**P**	**PM2, PM5, PM1_STRONG, PP2, PP5, PS4_MODERATE**	**CM900199**	-	**0.0001**	**unknown**	**no**	**positive (triplex)**	**m**	**34.89**
ADRP 521	*ADGRV1*	NM_032119.3	c.14432C>A	p.(Pro4811Gln)	NC_000005.9:g.90087078C>A	het	AR	VUS	PM2	-	-	0.0003	unknown	no	positive (triplex)	m	34.89
ADRP 521	*ADGRV1*	NM_032119.3	c.4854_4869dup	p.(Val1624Trpfs*3)	NC_000005.9:g.89968464_89968479dup	het	AR	P	PM2, PVS1, PP5	-	-	-	unknown	no	positive (triplex)	m	34.89
ARRP 110	***CRB1***	**NM_201253.2**	**c.2843G>A**	**p.(Cys948Tyr)**	**NC_000001.10:g.197403836G>A**	**hom**	**AR**	**P**	**PP3, PM2, PM5, PP2, PP5_STRONG, PS4_MODERATE**	**CM992152**	**39614**	**0.0002**	**unknown**	**Panel**	**positive (duplex)**	**m**	**53.30**
ARRP 110	*GUCA1B*	NM_002098.5	c.560G>A	p.(Ser187Asn)	NC_000006.11:g.42152596C>T	het	AD	VUS	PM2	-	-	0.0001	unknown	Panel	positive (duplex)	m	53.30
ARRP 125	***CACNA1F***	**NM_005183.2**	**c.26-2A>G**	**p.(?)**	**NC_000023.10:g.49088391T>C**	**hemi**	**XLR**	**LP**	**PM2, PVS1**	-	-	-	**unknown**	**MIP**	**positive (duplex)**	**m**	**51.71**
ARRP 125	*RP11*	NM_178857.5	c.2107C>T	p.(Arg703*)	NC_000008.10:g.10469501G>A	hom	AR+AD	P	PM2, PVS1_STRONG, PPS_STRONG	-	-	0.0001	unknown	MIP	positive (duplex)	m	51.71
ARRP 195	***ABCA4***	**NM_000350.2**	**c.2588G>C**	**p.(Gly863Ala)**	**NC_000001.10:g.94517254C>G**	**het**	**AR**	**P**	**PP3, PM2, PP2, PPS_STRONG, PS4_MODERATE**	**CM970003&CS024003**	-	**0.0044**	**unknown**	**Panel**	**positive (duplex)**	**m**	**50.86**
ARRP 195	***ABCA4***	**NM_000350.2**	**c.5603A>T**	**p.(Asn1868Ile)**	**NC_000001.10:g.94476467T>A**	**hom**	**AR**	**VUS**	**PP2, PP5, BA1, BS2**	**CM015091**	-	**0.0426**	**unknown**	**Panel**	**positive (duplex)**	**m**	**50.86**
ARRP 195	*PRCD*	NM_001077620.2	c.107G>A	p.(Gly36Asp)	NC_000017.10:g.74536619G>A	het	AR	VUS	PM2, BP4	-	-	0.0001	unknown	Panel	positive (duplex)	m	50.86
ARRP 245	***TMEM126B***	**NM_018480.4**	**c.635G>T**	**p.(Gly212Val)**	**NC_000011.9:g.85347215G>T**	**hom**	**AR**	**LP**	**PM2, PPS_STRONG**	**CM167900**	-	**0.001**	**unknown**	**Panel**	**negative**	**m**	**54.29**
ARRP 262	***RHO***	**NM_000539.3**	**c.14A>C**	**p.(Glu5Ala)**	**NC_000003.11:g.129247590A>C**	**het**	**AR+AD**	**LP**	**PP3, PM2, PM1, PP2**	-	-	-	**unknown**	**MIP**	**negative**	**f**	**35.78**
ARRP 274	***EYS***	**NM_001142800.1**	**c.1673G>A**	**p.(Trp558*)**	**NC_000006.11:g.66044966C>T**	**het**	**AR**	**P**	**PM2, PVS1, PP5**	**CM103594**	**522342**	**0.0001**	**unknown**	**Panel**	**positive (duplex)**	**f**	**51.77**
ARRP 274	***EYS***	**NM_001142800.1**	**c.6174T>G**	**p.(Tyr2058*)**	**NC_000006.11:g.65016880A>C**	**het**	**AR**	**P**	**PM2, PVS1, PP5**	**CM2117614**	-	-	**unknown**	**Panel**	**positive (duplex)**	**f**	**51.77**
ARRP 286	***USH2A***	**NM_206933.2**	**c.12574C>T**	**p.(Arg4192Cys)**	**NC_000001.10:g.215848679G>A**	**het**	**AR**	**P**	**PS1, PM2, PM5, PS4_MODERATE**	**CM137591**	**281818**	**0.0001**	**unknown**	**Panel**	**positive (triplex)**	**m**	**29.99**
ARRP 286	***USH2A***	**NM_206933.2**	**c.176G>A**	**p.(Gly59Glu)**	**NC_000001.10:g.216595503C>T**	**het**	**AR**	**LP**	**PM2, PPS_STRONG**	**CM140500**	-	**0.000007**	**unknown**	**Panel**	**positive (triplex)**	**m**	**29.99**
ARRP 347	***FLVCR1***	**NM_014053.3**	**c.1092+5G>A**	**p.(?)**	**NC_000001.10:g.213056785G>A**	**het**	**AR**	**P**	**PP3, PM2, PPS_STRONG**	**CS140551**	**374768**	**0.0003**	**unknown**	**Panel**	**positive (duplex)**	**f**	**39.60**
ARRP 347	***FLVCR1***	**NM_014

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ARRP 490	***EYS***	**NM_001142800.1**	**c.6191+2T>C**	**p.(?)**	**NC_000006.11:g.65016861A>G**	**het**	**AR**	**P**	**PM2, PVS1, PP5**	**-**	**865983**	**-**	**unknown**	**no**	**unknown**	**m**	**39.14**
ARRP 491	***PRPF31***	**NM_015629.3**	**c.239-620_1375-747del**	**p.(?)**	**NC_000019.9:g.54624619_54633991del**	**het**	**AD**	**P**	**PM2, PVS1, PP5**	**na**	**-**	**na**	**unknown**	**no**	**positive (duplex)**	**f**	**39.26**
ARRP 491	*RP1*	NM_006269.1	c.6065A>G	p.(Lys2022Arg)	NC_000008.10:g.55542507A>G	het	AD+AR	VUS	PM2	-	-	-	unknown	no	positive (duplex)	f	39.26
BBS 33	***MKKS***	**NM_170784.2**	**c.110A>G**	**p.(Tyr37Cys)**	**NC_000020.10:g.10394053T>C**	**hom**	**AR**	**P**	**PP3, PM2, PP5_STRONG**	**CM001240**	**5309**	**0.0001**	**unknown**	**no**	**negative**	**m**	**46.99**
BBS 35	***BBS2***	**NM_031885.3**	**c.1895G>C**	**p.(Arg632Pro)**	**NC_000016.9:g.56530894C>G**	**het**	**AR**	**LP**	**PP3, PM2, PP5_STRONG**	**CM016198**	**-**	**0.0001**	**unknown**	**no**	**negative**	**m**	**52.09**
BBS 35	***BBS2***	**NM_031885.3**	**c.661del**	**p.(Leu221Phefs*25)**	**NC_000016.9:g.56540090del**	**het**	**AR**	**P**	**PM2, PVS1, PP5**	**CD101918**	**-**	**0.000025**	**unknown**	**no**	**negative**	**m**	**52.09**
BBS 70	***BBS7***	**NM_176824.2**	**c.712_715del**	**p.(Arg238Glufs*59)**	**NC_000004.11:g.122775864_122775867del**	**het**	**AR**	**P**	**PM2, PVS1_MODERATE, PP5_STRONG, PS4_MODERATE**	**CD093973**	**281626**	**0.00005**	**unknown**	**no**	**negative**	**f**	**54.50**
BBS 70	***BBS7***	**NM_176824.2**	**c.968A>G**	**p.(His323Arg)**	**NC_000004.11:g.122768628T>C**	**het**	**AR**	**LP**	**PM2, PP5_STRONG**	**CM030645**	**3015**	**0.00004**	**unknown**	**no**	**negative**	**f**	**54.50**
BBS 72	***BBS10***	**NM_024685.3**	**c.145C>T**	**p.(Arg49Trp)**	**NC_000012.11:g.76741994G>A**	**hom**	**AR**	**LP**	**PM2, PP5_STRONG**	**CM063864**	**225010**	**0.0001**	**unknown**	**no**	**negative**	**f**	**51.81**
BCM 308-1	***OPN1LW***	**NM_020061.4**	**c.532A>G**	**p.(Ile178Val)**	**NC_000023.10:g.153418535A>G**	**hemi**	**XLR**	**B**	**BS1, BS2, BP4, BP6_STRONG**	**CM890024**	**-**	**0.0214**	**unknown**	**no**	**positive (duplex)**	**m**	**46.21**
BCM 308-1	***OPN1LW***	**NM_020061.4**	**c.538T>G**	**p.(Ser180Ala)**	**NC_000023.10:g.153418541T>G**	**hemi**	**XLR**	**B**	**BA1, BS2, BP6**	**CM045803**	**-**	**0.2244**	**unknown**	**no**	**positive (duplex)**	**m**	**46.21**
BCM 308-1	***OPN1MW***	**NM_000513.1**	**c.532A>G**	**p.(Ile178Val)**	**NC_000023.10:g.153455665A>G**	**hemi**	**XLR**	**B**	**BA1, BS1, BS2, BP4**	**CM890024**	**-**	**0.0214**	**unknown**	**no**	**positive (duplex)**	**m**	**46.21**
BCM 308-2	***OPN1LW***	**NM_020061.4**	**c.532A>G**	**p.(Ile178Val)**	**NC_000023.10:g.153418535A>G**	**hemi**	**XLR**	**B**	**BS1, BS2, BP4, BP6_STRONG**	**CM890024**	**-**	**0.0214**	**unknown**	**no**	**positive (duplex)**	**m**	**44.94**
BCM 308-2	***OPN1LW***	**NM_020061.4**	**c.538T>G**	**p.(Ser180Ala)**	**NC_000023.10:g.153418541T>G**	**hemi**	**XLR**	**B**	**BA1, BS2, BP6**	**CM045803**	**-**	**0.2244**	**unknown**	**no**	**positive (duplex)**	**m**	**44.94**
BCM 323	***OPN1LW***	**NM_020061.4**	**c.607T>C**	**p.(Cys203Arg)**	**NC_000023.10:g.153420077T>C**	**hemi**	**XLR**	**LP**	**PM2, PP5_STRONG**	**CM951111**	**-**	**-**	**unknown**	**single genes**	**positive (duplex)**	**m**	**52.83**
BCM 342	***OPN1LW***	**NM_020061.4**	**na**	**na**	**NC_000023.10:g.153400836_153408770del**	**hemi**	**XLR**	**LP**	**PM2, PVS1**	**na**	**-**	**na**	**unknown**	**no**	**unknown**	**m**	**52.89**
BCM 346	***OPN1LW***	**NM_020061.4**	**c.532A>G**	**p.(Ile178Val)**	**NC_000023.10:g.153418535A>G**	**hemi**	**XLR**	**B**	**BS1, BS2, BP4, BP6_STRONG**	**CM890024**	**-**	**0.0214**	**unknown**	**single genes**	**positive (duplex)**	**m**	**47.39**
BCM 346	***OPN1LW***	**NM_020061.4**	**c.538T>G**	**p.(Ser180Ala)**	**NC_000023.10:g.153418541T>G**	**hemi**	**XLR**	**B**	**BA1, BS2, BP6**	**CM045803**	**-**	**0.2244**	**unknown**	**single genes**	**positive (duplex)**	**m**	**47.39**
BCM 354	***OPN1LW***	**NM_020061.4**	**c.607T>C**	**p.(Cys203Arg)**	**NC_000023.10:g.153420077T>C**	**hemi**	**XLR**	**LP**	**PM2, PP5_STRONG**	**CM951111**	**-**	**-**	**unknown**	**single genes**	**positive (duplex)**	**m**	**50.89**
BCM 371	***OPN1LW***	**NM_020061.4**	**c.607T>C**	**p.(Cys203Arg)**	**NC_000023.10:g.153420077T>C**	**hemi**	**XLR**	**LP**	**PM2, PP5_STRONG**	**CM951111**	**-**	**-**	**unknown**	**no**	**negative**	**m**	**37.00**
BCM 379	***OPN1LW***	**NM_020061.4**	**na**	**na**	**NC_000023.10:g.153400711_153414443del**	**het**	**XLR**	**LP**	**PM2, PVS1**	**na**	**-**	**-**	**unknown**	**no**	**positive (duplex)**	**f**	**47.98**
BVMD 48	***BEST1***	**NM_004183.3**	**c.703G>T**	**p.(Val235Leu)**	**NC_000011.9:g.61724925G>T**	**het**	**AD+AR**	**P**	**PP3, PM2, PM5_SUPPORTING, PM1, PP2, PP5_STRONG**	**CM1517494**	**-**	**-**	**unknown**	**no**	**positive (multiplex)**	**f**	**51.11**
BVMD 65	***PRPH2***	**NM_000322.4**	**c.948G>A**	**p.(Trp316*)**	**NC_000006.11:g.42666126C>T**	**het**	**AD**	**P**	**PM2, PVS1_STRONG, PP5_STRONG**	**CM2036762**	**-**	**-**	**unknown**	**Panel**	**positive (triplex)**	**f**	**52.68**
BVMD 98	***BEST1***	**NM_004183.3**	**c.693T>A**	**p.(Ser231Arg)**	**NC_000011.9:g.61724915T>A**	**het**	**AD+AR**	**P**	**PS1, PP3, PM2, PM1, PP2, PP5**	**-**	**-**	**-**	**unknown**	**no**	**negative**	**f**	**52.81**
BVMD 103	***BEST1***	**NM_004183.3**	**c.684C>G**	**p.(Asp228Glu)**	**NC_000011.9:g.61724906C>G**	**het**	**AD+AR**	**P**	**PP3, PM2, PM5_SUPPORTING, PM1, PP2, PP5_STRONG**	**CM183768**	**522450**	**0.000004**	**unknown**	**no**	**positive (duplex)**	**m**	**57.91**
BVMD 103	*IMPG2*	NM_016247.3	c.3272A>C	p.(His1091Pro)	NC_000003.11:g.100949951T>G	het	AD	VUS	PM2	-	-	0.000004	unknown	no	positive (duplex)	m	57.91
BVMD 104	***BEST1***	**NM_004183.3**	**c.728C>T**	**p.(Ala243Val)**	**NC_000011.9:g.61725631C>T**	**het**	**AD+AR**	**P**	**PP3, PM2, PM5, PM1_SUPPORTING, PP2, PP5_STRONG**	**CM000841**	**-**	**-**	**unknown**	**no**	**positive (multiplex)**	**f**	**50.61**
BVMD 105	***BEST1***	**NM_004183.3**	**c.297C>A**	**p.(Asn99Lys)**	**NC_000011.9:g.61723239C>A**	**het**	**AD+AR**	**P**	**PP3, PM2, PM5_SUPPORTING, PM1, PP2, PP5_STRONG**	**CM000837**	**-**	**-**	**unknown**	**no**	**negative**	**f**	**56.88**
BVMD 105	***BEST1***	**NM_004183.3**	**c.422G>A**	**p.(Arg141His)**	**NC_000011.9:g.61723364G>A**	**het**	**AD+AR**	**P**	**PP3, PM2, PM5, PM1, PP2, PP5_STRONG**	**CM001382**	**2740**	**0.0006**	**unknown**	**no**	**negative**	**f**	**56.88**
BVMD 106	***BEST1***	**NM_004183.3**	**c.182del**	**p.(Met61Serfs*5)**	**NC_000011.9:g.61722608del**	**hom**	**AR+AD**	**LP**	**PM2, PVS1_MODERATE, PP5_STRONG**	**-**	**987408**	**-**	**unknown**	**no**	**positive (duplex)**	**f**	**52.14**
BVMD 110	***BEST1***	**NM_004183.3**	**c.665G>T**	**p.(Gly222Val)**	**NC_000011.9:g.61724887G>T**	**het**	**AR+AD**	**LP**	**PP3, PM2, PP2, PP5_STRONG**	**CM003966**	**99739**	**-**	**maternal**	**no**	**negative**	**m**	**39.72**
BVMD 114-1	***BEST1***	**NM_004183.3**	**c.1315C>T**	**p.(Gln439*)**	**NC_000011.9:g.61729941C>T**	**het**	**AR+AD**	**P**	**PM2, PVS1, PP5**	**CM2120844**	**813025**	**-**	**unknown**	**no**	**positive (duplex)**	**m**	**36.80**
BVMD 114-1	***BEST1***	**NM_004183.3**	**c.910_912del**	**p.(Asp304del)**	**NC_000011.9:g.61727012_61727014del**	**het**	**AR+AD**	**LP**	**PM2, PM4, PM5_SUPPORTING, PM1**	**CD004449**	**-**	**-**	**unknown**	**no**	**positive (duplex)**	**m**	**36.80**
BVMD 114-2	***BEST1***	**NM_004183.3**	**c.910_912del**	**p.(Asp304del)**	**NC_000011.9:g.61727012_61727014del**	**het**	**AR+AD**	**LP**	**PM2, PM4, PM5_SUPPORTING, PM1**	**CD004449**	**-**	**-**	**unknown**	**no**	**positive (duplex)**	**f**	**42.52**
BVMD 116	***BEST1***	**NM_004183.3**	**c.874G>A**	**p.(Glu292Lys)**	**NC_000011.9:g.61726976G>A**	**het**	**AR+AD**	**LP**	**PP3, PM2, PP2, PP5_STRONG**	**CM094327**	**-**	**-**	**unknown**	**no**	**negative**	**f**	**38.85**
BVMD 117	***IMPG2***	**NM_016247.3**	**c.676G>T**	**p.(Glu226*)**	**NC_000003.11:g.100992577C>A**	**het**	**AD**	**P**	**PM2, PVS1, PP5**	**CM179467**	**1275777**	**-**	**unknown**	**no**	**negative**	**f**	**39.73**
CACD 25	***ABCA4***	**NM_000350.2**	**c.5196+1137G>A**	**p.(?)**	**NC_000001.10:g.94484001C>T**	**het**	**AR**	**P**	**PM2, PP5_STRONG, BP7, PS4_MODERATE**	**CS1313969**	**-**	**0.000096**	**unknown**	**no**	**positive (duplex)**	**m**	**56.09**
CACD 25	***ABCA4***	**NM_000350.2**	**c.5311G>A**	**p.(Gly1771Arg)**	**NC_000001.10:g.94481296C>T**	**het**	**AR**	**LP**	**PP3, PM2, PM1_SUPPORTING, PP2, PP5**	**CM160499**	**-**	**0.000004**	**unknown**	**no**	**positive (duplex)**	**m**	**56.09**
CACD 84	***ABCA4***	**NM_000350.2**	**c.1622T>C**	**p.(Leu541Pro)**	**NC_000001.10:g.94528806A>G**	**het**	**AR**	**P**	**PP3, PM2, PP2, PP5_STRONG, PS4_MODERATE**	**CM990022**	**7901**	**0.0003**	**not transmitted to son**	**no**	**negative**	**f**	**54.89**
CACD 84	***ABCA4***	**NM_000350.2**	**c.3113C>T**	**p.(Ala1038Val)**	**NC_000001.10:g.94508969G>A**	**het**	**AR**	**P**	**PM2, PM1_SUPPORTING, PP2, PP5_STRONG, PS4_MODERATE**	**CM970006**	**7901**	**0.0024**	**not transmitted to son**	**no**	**negative**	**f**	**54.89**
CACD 84	***ABCA4***	**NM_000350.2**	**c.4237C>A**	**p.(Gln1413Lys)**	**NC_000001.10:g.94496568G>T**	**het**	**AR**	**LP**	**PP3, PM2, PM1_SUPPORTING, PP2, PM3**	**-**	**-**	**-**	**transmitted to son**	**no**	**negative**	**f**	**54.89**
CACD 85	***ABCA4***	**NM_000350.2**	**c.4457C>T**	**p.(Pro1486Leu)**	**NC_000001.10:g.94495083G>A**	**het**	**AR**	**P**	**PP3, PM2, PM1_SUPPORTING, PP2, PP5_STRONG**	**CM990054**	**99283**	**0.0001**	**unknown**	**no**	**negative**	**f**	**40.22**
CACD 85	***ABCA4***	**NM_000350.2**	**c.5175dup**	**p.(Thr1726Aspfs*61)**	**NC_000001.10:g.94485159dup**	**het**	**AR**	**P**	**PM2, PVS1, PP5**	**CI144136**	**-**	**0.0001**	**unknown**	**no**	**negative**	**f**	**40.22**
CACD 85	***ABCA4***	**NM_000350.2**	**c.5603A>T**	**p.(Asn1868Ile)**	**NC_000001.10:g.94476467T>A**	**het**	**AR**	**VUS**	**PP2, PP5, BA1, BS2**	**CM015091**	**-**	**0.0426**	**unknown**	**no**	**negative**	**f**	**40.22**
CD 1	***GUCA1A***	**NM_000409.3**	**c.451C>T**	**p.(Leu151Phe)**	**NC_000006.11:g.42146986C>T**	**het**	**AD**	**LP**	**PM2, PP2, PP5_STRONG**	**CM053920**	**-**	**0.000007**	**unknown**	**single genes**	**positive (multiplex)**	**m**	**38.57**
CD 1	*COL18A1*	NM_030582.3	c.3206dup	p.(Gly1072Argfs*9)	NC_000021.8:g.46917558dup	het	AR	P	PM2, PVS1, PP5	CI092465	-	0.0001	unknown	single genes	positive (multiplex)	m	38.57
CD 1	*TGFBI*	NM_000358.2	c.371G>A	p.(Arg124His)	NC_000005.9:g.135382096G>A	het	AD	LP	PP3, PM2, PM5, PP5	CM971472	-	0.0001	unknown	single genes	positive (multiplex)	m	38.57
CD 391	***CNGA3***	**NM_001298.2**	**c.1694C>T**	**p.(Thr565Met)**	**NC_000002.11:g.99013327C>T**	**hom**	**AR**	**P**	**PP3, PM2, PM1, PP2, PP5_STRONG**	**CM014558**	**867082**	**0.0003**	**unknown**	**no**	**positive (duplex)**	**m**	**49.04**
CD 395	***KCNV2***	**NM_133497.3**	**c.1211T>C**	**p.(Leu404Pro)**	**NC_000009.11:g.2718950T>C**	**het**	**AR**	**LP**	**PP3, PM2, PP5_STRONG**	**CM1110039**	**-**	**-**	**unknown**	**single genes**	**positive (duplex)**	**m**	**47.09**
CD 395	***KCNV2***	**NM_133497.2**	**na**	**na**	**NC_000009.11:g.2667638_2747340del**	**het**	**AR**	**LP**	**PM2, PVS1**	**na**	**-**	**na**	**unknown**	**single genes**	**positive (duplex)**	**m**	**47.09**
CD 419	***CRX***	**NM_000554.4**	**c.122G>A**	**p.(Arg41Gln)**	**NC_000019.9:g.48339521G>A**	**het**	**AD**	**LP**	**PP3, PM2, PP5_STRONG**	**CM970396**	**-**	**0.0001**	**unknown**	**Panel**	**negative**	**f**	**48.10**
CD 557	***ABCA4***	**NM_000350.2**	**c.4234C>T**	**p.(Gln1412*)**	**NC_000001.10:g.94496571G>A**	**het**	**AR**	**P**	**PM2, PVS1, PP5**	**CM990049**	**-**	**0.0001**	**unknown**	**Panel**	**unknown**	**f**	**52.11**
CD 557	***ABCA4***	**NM_000350.2**	**c.4253+43G>A**	**p.(?)**	**NC_000001.10:g.94496509C>T**	**het**	**AR**	**LP**	**PM2, PP5_STRONG, BP7**	**CS193151**	**-**	**0.0047**	**unknown**	**Panel**	**unknown**	**f**	**52.11**
CD 609	***CLN3***	**NM_001042432.1**	**c.461-280_677+382del**	**p.(?)**	**NC_000016.9:g.28497286_28498251del**	**hom**	**AR**	**LP**	**PM2, PVS1**	**na**	**-**	**na**	**paternal/maternal**	**no**	**negative**	**f**	**49.11**
CD 619	***GUCY2D***	**NM_000180.3**	**c.2513G>A**	**p.(Arg838His)**	**NC_000017.10:g.7918019G>A**	**het**	**AR+AD**	**P**	**PS1_MODERATE, PP3, PM2, PM5, PM1, PP2, PP5_STRONG, PS4_MODERATE**	**CM012606**	**-**	**-**	**unknown**	**single genes**	**negative**	**m**	**54.64**
CD 620	***ABCA4***	**NM_000350.2**	**c.1988G>A**	**p.(Trp663*)**	**NC_000001.10:g.94526265C>T**	**het**	**AR**	**P**	**PM2, PVS1, PP5**	**CM003370**	**372289**	**-**	**unknown**	**no**	**negative**	**f**	**47.38**
CD 620	***ABCA4***	**NM_000350.2**	**c.4918C>T**	**p.(Arg1640Trp)**	**NC_000001.10:g.94486896G>A**	**het**	**AR**	**P**	**PS1_MODERATE, PP3, PM2, PM5, PM1_SUPPORTING, PP2, PP5_STRONG**	**CM983728**	**-**	**0.0001**	**unknown**	**no**	**negative**	**f**	**47.38**
CD 621	***CRX***	**NM_000554.4**	**na**	**na**	**NC_000019.9:g.48340644_48406926del**	**het**	**AD**	**LP**	**PM2, PVS1**	**na**	**-**	**na**	**unknown**	**no**	**positive (duplex)**	**f**	**29.77**
CD 624	***RPGR***	**NM_001034853.1**	**c.3081_3082del**	**p.(Glu1028Glyfs*50)**	**NC_000023.10:g.38145172_38145173del**	**hemi**	**XLD+XLR**	**LP**	**PM2, PVS1_STRONG, PP5**	**-**	**-**	**-**	**unknown**	**no**	**negative**	**m**	**51.07**
CD 626	***GUCA1A***	**NM_000409.3**	**c.250C>T**	**p.(Leu84Phe)**	**NC_000006.11:g.42146066C>T**	**het**	**AD**	**LP**	**PM2, PP2, PP5_STRONG**	**CM138778**	**-**	**-**	**unknown**	**no**	**positive (multiplex)**	**f**	**58.16**
CD 626	*EYS*	NM_001142800.1	c.9418C>T	p.(Gln3140*)	NC_000006.11:g.64430509G>A	het	AR	LP	PM2, PVS1_STRONG	-	-	-	unknown	no	positive (multiplex)	f	58.16
CD 628	***KCNV2***	**NM_133497.3**	**c.741_742delinsAC**	**p.(Trp247*)**	**NC_000009.11:g.2718480_2718481delinsAC**	**hom**	**AR**	**LP**	**PM2, PVS1**	**-**	**1184889**	**-**	**unknown**	**no**	**negative**	**m**	**40.36**
CD 630	***RPGR***	**NM_001034853.1**	**c.3070G>T**	**p.(Glu1024*)**	**NC_000023.10:g.38145182C>A**	**hemi**	**XLD+XLR**	**P**	**PM2, PVS1, PP5**	**CM144449**	**1275798**	**-**	**unknown**	**no**	**negative**	**m**	**41.24**
CD 633	***CRX***	**NM_000554.4**	**c.-35-1771_101-707del**	**p.(?)**	**NC_000019.9:g.48335895_48338793del**	**het**	**AD**	**LP**	**PM2, PVS1**	**na**	**-**	**na**	**unknown**	**no**	**unknown**	**f**	**38.34**
CHM 15	***CHM***	**NM_000390.2**	**c.116+1G>A**	**p.(?)**	**NC_000023.10:g.85282494C>T**	**het**	**XLD**	**P**	**PM2, PVS1, PP5**	**CS032061**	**-**	**-**	**unknown**	**no**	**positive (duplex)**	**f**	**52.31**
CHM 27-1	***CHM***	**NM_000390.2**	**c.1644G>A**	**p.(Trp548*)**	**NC_000023.10:g.85128183C>T**	**hemi**	**XLD**	**P**	**PM2, PVS1, PP5**	**CM031953**	**-**	**-**	**unknown**	**no**	**positive (multiplex)**	**m**	**51.40**
CHM 27-2	***CHM***	**NM_000390.2**	**c.1644G>A**	**p.(Trp548*)**	**NC_000023.10:g.85128183C>T**	**hemi**	**XLD**	**P**	**PM2, PVS1, PP5**	**CM031953**	**-**	**-**	**unknown**	**no**	**positive (multiplex)**	**m**	**58.17**
CHM 109	***CHM***	**NM_000390.2**	**c.315-4587T>A**	**p.(?)**	**NC_000023.10:g.85223644A>T**	**hemi**	**XLD**	**LP**	**PM2, PP5_STRONG**	**CS032064**	**-**	**-**	**unknown**	**no**	**negative**	**m**	**45.93**
CHM 111	***CHM***	**NM_000390.2**	**c.941-1G>T**	**p.(?)**	**NC_000023.10:g.85211384C>A**	**hemi**	**XLD**	**P**	**PM2, PVS1, PP5**	**CS1515666**	**-**	**-**	**unknown**	**no**	**positive (duplex)**	**m**	**33.84**
CHM 111	*GUCY2D*	NM_000180.3	c.7919dup	p.(Pro1074Thrfs*58)	NC_000017.10:g.7919603dup	het	AR+AD	LP	PM2, PVS1_STRONG, PP5	-	-	-	unknown	no	positive (duplex)	m	33.84
CHM 112	***CHM***	**NM_000390.2**	**c.808C>T**	**p.(Arg270*)**	**NC_000023.10:g.85213877G>A**	**hemi**	**XLD**	**P**	**PM2, PVS1, PP5**	**CM983732**	**143078**	**-**	**unknown**	**no**	**positive (duplex)**	**m**	**61.53**
CHM 119	***CHM***	**NM_000390.2**	**c.703-121_1166+9561del**	**p.(?)**	**NC_000023.10:g.85201597_85214103del**	**hemi**	**XLD**	**LP**	**PM2, PVS1**	**na**	**-**	**na**	**unknown**	**no**	**positive (triplex)**	**m**	**43.39**
CHM 120	***CHM***	**NM_000390.2**	**c.262_263dup**	**p.(Ser89Leufs*38)**	**NC_000023.10:g.85233824_85233825dup**	**hemi**	**XLD**	**P**	**PM2, PVS1, PP5**	**-**	**-**	**-**	**unknown**	**no**	**positive (duplex)**	**m**	**54.92**
CHM 122	***CHM***	**NM_000390.2**	**na**	**na**	**NC_000023.10:g.84785132_86225549del**	**hemi**	**XLD**	**LP**	**PM2, PVS1**	**na**	**-**	**na**	**unknown**	**no**	**negative**	**m**	**48.53**
CHM 123	***CHM***	**NM_000390.2**	**na**	**na**	**NC_000023.10:g.84646897_85634486del**	**hemi**	**XLD**	**LP**	**PM2, PVS1**	**na**	**-**	**na**	**unknown**	**no**	**positive (duplex)**	**m**	**41.16**
CHM 126	***CHM***	**NM_000390.2**	**c.525_526del**	**p.(Glu177Lysfs*6)**	**NC_000023.10:g.85218846_85218847del**	**hemi**	**XLD**	**P**	**PM2, PVS1, PP5**	**CD941644**	**-**	**-**	**unknown**	**Panel**	**positive (duplex)**	**m**	**45.34**
CHM 126	*ADAM9*	NM_003816.2	c.2245C>T	p.(Gln749*)	NC_000008.10:g.38948812C>T	het	AR	LP	PM2, PVS1	-	-	-	unknown	Panel	positive (duplex)	m	45.34
CHM 129	***CHM***	**NM_000390.2**	**c.1370T>C**	**p.(Leu457Pro)**	**NC_000023.10:g.85155694A>G**	**hemi**	**XLD**	**LP**	**PP3, PM2, PP5_STRONG**	**CM166157**	**1275780**	**-**	**unknown**	**no**	**negative**	**m**	**42.13**
CRD 102	***AIPL1***	**NM_014336.3**	**c.1010_1011del**	**p.(Glu337Alafs*70)**	**NC_000017.10:g.6328926_6328927del**	**hom**	**AR**	**LP**	**PM2, PVS1**	**CD001471**	**-**	**-**	**unknown**	**single genes**	**negative**	**m**	**40.52**
CRD 337	***ABCA4***	**NM_000350.2**	**c.4462T>C**	**p.(Cys1488Arg)**	**NC_000001.10:g.94495078A>G**	**het**	**AR**	**P**	**PP3, PM2, PM5, PM1_SUPPORTING, PP2, PP5_STRONG**	**CM990055**	**99284**	**0.0001**	**unknown**	**single genes**	**positive (duplex)**	**m**	**40.05**
CRD 337	***ABCA4***	**NM_000350.2**	**c.5917del**	**p.(Val1973*)**	**NC_000001.10:g.94473278del**	**het**	**AR**	**P**	**PM2, PVS1, PP5, PS4**	**CD003502**	**-**	**0.0001**	**unknown**	**single genes**	**positive (duplex)**	**m**	**40.05**
CRD 464	***PRPH2***	**NM_000322.4**	**c.760del**	**p.(Leu254Cysfs*2)**	**NC_000006.11:g.42672173del**	**het**	**AD**	**P**	**PM2, PVS1, PP5**	**-**	**-**	**-**	**unknown**	**no**	**unknown**	**f**	**49.58**
CRD 658	***CERKL***	**NM_201548.4**	**c.1363C>T**	**p.(Gln455*)**	**NC_000002.11:g.182409429G>A**	**hom**	**AR**	**P**	**PM2, PVS1, PP5**	**-**	**-**	**0.0001**	**unknown**	**single genes**	**negative**	**m**	**54.14**
CRD 668	***POC1B***	**NM_172240.2**	**c.1340C>G**	**p.(Ser447*)**	**NC_000012.11:g.89815027G>C**	**het**	**AR**	**P**	**PM2, PVS1, PM3**	**-**	**-**	**-**	**unknown**	**Panel**	**negative**	**m**	**42.03**
CRD 668	***POC1B***	**NM_172240.2**	**c.561-3T>C**	**p.(Phe188Sfs*6)**	**NC_000012.11:g.89865509A>G**	**het**	**AR**	**LP**	**PM2, PM3, PS3, BP4**	**-**	**-**	**-**	**unknown**	**Panel**	**negative**	**m**	**42.03**
CRD 671	***SCLT1***	**NM_144643.2**	**c.1294-2A>G**	**p.(?)**	**NC_000004.11:g.129867309T>C**	**het**	**AR**	**LP**	**PM2, PVS1, PM3, PS3**	**-**	**-**	**-**	**paternal**	**Panel**	**negative**	**m**	**44.69**
CRD 671	***SCLT1***	**NM_144643.2**	**c.2005-1052_*363del**	**p.(?)**	**NC_000004.11:g.129805272_129806749del**	**het**	**AR**	**LP**	**PM2, PVS1**	**na**	**-**	**na**	**maternal**	**Panel**	**negative**	**m**	**44.69**
CRD 714	***PRPH2***	**NM_000322.4**	**c.664T>C**	**p.(Cys222Arg)**	**NC_000006.11:g.42672267A>G**	**het**	**AD**	**LP**	**PP3, PM2, PM5, PM1, PP2, PP5**	**CM175541**	**-**	**-**	**unknown**	**no**	**negative**	**m**	**47.30**
CRD 766	***POC1B***	**NM_172240.2**	**c.1331_1332dup**	**p.(Thr445Argfs*10)**	**NC_000012.11:g.89818938_89818939dup**	**hom**	**AR**	**LP**	**PM2, PVS1_MODERATE, PP5_STRONG**	**-**	**987339**	**-**	**unknown**	**no**	**negative**	**f**	**47.66**
CRD 770-1	***ABCA4***	**NM_000350.2**	**c.4540-2A>G**	**p.(?)**	**NC_000001.10:g.94490606T>C**	**het**	**AR**	**P**	**PM2, PVS1, PP5**	**CS033103**	**92869**	**0.000004**	**unknown**	**single genes**	**positive (duplex)**	**f**	**44.93**
CRD 770-1	***ABCA4***	**NM_000350.2**	**c.6229C>T**	**p.(Arg2077Trp)**	**NC_000001.10:g.94467467G>A**	**het**	**AR**	**P**	**PP3, PM2, PM5, PM1, PP2, PP5_STRONG, PS4_MODERATE**	**CM970021**	**99438**	**0.0001**	**unknown**	**single genes**	**positive (duplex)**	**f**	**44.93**
CRD 770-2	***ABCA4***	**NM_000350.2**	**c.4540-2A>G**	**p.(?)**	**NC_000001.10:g.94490606T>C**	**het**	**AR**	**P**	**PM2, PVS1, PP5**	**CS033103**	**92869**	**0.000004**	**unknown**	**single genes**	**positive (duplex)**	**m**	**45.65**
CRD 770-2	***ABCA4***	**NM_000350.2**	**c.6229C>T**	**p.(Arg2077Trp)**	**NC_000001.10:g.94467467G>A**	**het**	**AR**	**P**	**PP3, PM2, PM5, PM1, PP2, PP5_STRONG, PS4_MODERATE**	**CM970021**	**99438**	**0.0001**	**unknown**	**single genes**	**positive (duplex)**	**m**	**45.65**
CRD 771	***ABCA4***	**NM_000350.2**	**c.4139C>T**	**p.(Pro1380Leu)**	**NC_000001.10:g.94496666G>A**	**het**	**AR**	**P**	**PP3, PM2, PM1_SUPPORTING, PP2, PP5_STRONG, PS4_MODERATE**	**CM990044**	**7904**	**0.0002**	**unknown**	**no**	**positive (duplex)**	**f**	**53.27**
CRD 771	***ABCA4***	**NM_000350.2**	**c.5196+1G>A**	**p.(?)**	**NC_000001.10:g.94485137C>T**	**het**	**AR**	**P**	**PM2, PVS1, PP5**	**CS971603**	**-**	**0.0001**	**unknown**	**no**	**positive (duplex)**	**f**	**53.27**
CRD 772	***GUCY2D***	**NM_000180.3**	**c.2513G>A**	**p.(Arg838His)**	**NC_000017.10:g.7918019G>A**	**het**	**AD+AR**	**P**	**PS1_MODERATE, PP3, PM2, PM5, PM1, PP2, PP5_STRONG, PS4_MODERATE**	**CM012606**	**-**	**-**	**unknown**	**no**	**positive (multiplex)**	**f**	**49.26**
CRD 773	***PCARE***	**NM_001029883.2**	**c.2704A>T**	**p.(Lys902*)**	**NC_000002.11:g.29294424T>A**	**het**	**AR**	**P**	**PM2, PVS1, PP5**	**-**	**987347**	**-**	**unknown**	**no**	**negative**	**m**	**48.81**
CRD 773	***PCARE***	**NM_001029883.2**	**c.776_777del**	**p.(Glu259Alafs*51)**	**NC_000002.11:g.29296355_29296356del**	**het**	**AR**	**P**	**PM2, PVS1, PP5**	**-**	**812239**	**-**	**unknown**	**no**	**negative**	**m**	**48.81**
CRD 773	*ABCA4*	NM_000350.2	c.5882G>A	p.(Gly1961Glu)	NC_000001.10:g.94473807C>T	het	AR	P	PP3, PM2, PM5, PP2, PP5_STRONG, PS4_MODERATE	CM970016	635154	0.0047	unknown	no	negative	m	48.81
CRD 776	***GUCY2D***	**NM_000180.3**	**c.2513G>A**	**p.(Arg838His)**	**NC_000017.10:g.7918019G>A**	**het**	**AD+AR**	**P**	**PS1_MODERATE, PP3, PM2, PM5, PM1, PP2, PP5_STRONG, PS4_MODERATE**	**CM012606**	**-**	**-**	**unknown**	**single genes**	**positive (multiplex)**	**f**	**46.83**
CRD 779	***ABCA4***	**NM_000350.2**	**c.1086T>A**	**p.(Tyr362*)**	**NC_000001.10:g.94546047A>T**	**het**	**AR**	**P**	**PM2, PVS1, PP5**	**CM003364**	**236078**	**-**	**unknown**	**no**	**negative**	**f**	**30.87**
CRD 779	***ABCA4***	**NM_000350.2**	**c.3364G>A**	**p.(Glu1122Lys)**	**NC_000001.10:g.94506923C>T**	**het**	**AR**	**P**	**PP3, PM2, PM1, PP2, PP5_STRONG, PS4_MODERATE**	**CM990041**	**-**	**0.00003**	**unknown**	**no**	**negative**	**f**	**30.87**
CRD 779	***ABCA4***	**NM_000350.2**	**c.5603A>T**	**p.(Asn1868Ile)**	**NC_000001.10:g.94476467T>A**	**het**	**AR**	**VUS**	**PP2, PP5, BA1, BS2**	**CM015091**	**-**	**0.0426**	**unknown**	**no**	**negative**	**f**	**30.87**
CRD 780	***IMPG2***	**NM_016247.3**	**na**	**na**	**NC_000003.11:g.101034946_101040925delinsCTTTGATCC**	**het**	**AD**	**LP**	**PM2, PVS1**	**na**	**-**	**na**	**unknown**	**no**	**positive (triplex)**	**f**	**55.66**
CRD 780	*IMPG2*	NM_016247.3	c.2717G>A	p.(Arg906Gln)	NC_000003.11:g.100962458C>T	het	AD	VUS	PM2	-	-	-	unknown	no	positive (triplex)	f	55.66
CRD 784	***ABCA4***	**NM_000350.2**	**c.4849-2A>G**	**p.(?)**	**NC_000001.10:g.94486967T>C**	**hom**	**AR**	**P**	**PM2, PVS1, PP5**	**-**	**986969**	**-**	**unknown**	**no**	**negative**	**m**	**46.03**
CRD 785	***ABCA4***	**NM_000350.2**	**

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LCA 161	*RPE65*	NM_000329.2	c.304G>T	p.(Glu102*)	NC_000001.10:g.68910508C>A	hom	AR	P	PM2, PVS1, PP5	CM994069	98863	0.0001	paternal/maternal	no	negative	f	64.42
LCA 163	*IQCB1*	NM_001023570.2	c.1518_1519del	p.(His506Glnfs*13)	NC_000003.11:g.121491454_121491455del	hom	AR	P	PM2, PVS1_STRONG, PP5_STRONG	CD050887	93469	0.0001	maternal/paternal	no	negative	f	35.90
LCA 174	*RPE65*	NM_000329.2	c.209T>C	p.(Phe70Ser)	NC_000001.10:g.68912429A>G	hom	AR	P	PP3, PM2, PM5_SUPPORTING, PM1, PP2, PP5_STRONG	CM1816029	-	-	paternal/maternal	no	unknown	m	59.36
LCA 179	*CEP290*	NM_025114.3	c.2368-831_2483+627del	p.(?)	NC_000012.11:g.88502219_88503792del	het	AR	LP	PM2, PVS1	na	-	na	unknown	no	unknown	m	47.09
LCA 179	*CEP290*	NM_025114.3	c.2991+1655A>G	p.(?)	NC_000012.11:g.88494960T>C	het	AR	LP	PM2, PP5_STRONG, BP7	CS064383	-	0.0001	unknown	no	unknown	m	47.09
LCA 181	*CEP290*	NM_025114.3	c.4159dup	p.(Ser1387Lysfs*4)	NC_000012.11:g.88481592dup	het	AR	P	PM2, PVS1, PP5	-	448961	-	unknown	no	negative	f	61.94
LCA 181	*CEP290*	NM_025114.3	c.4661_4663del	p.(Glu1554del)	NC_000012.11:g.88478410_88478412del	het	AR	LP	PM2, PM4, PP5_STRONG	CD072352	982524	0.0001	unknown	no	negative	f	61.94
LCA 181	*USP45*	NM_001080481.1	c.9del	p.(Asp5Ilefs*30)	NC_000006.11:g.99958088del	het	AR	LP	PM2, PVS1	-	-	0.0001	unknown	no	negative	f	61.94
LHON 1201	*MT-ND4*	NC_012920.1	m.11778G>A	p.(Arg340His)	NC_012920.1:m.11778G>A (homoplasmic, 99.8% of reads)	na	MT	P	PP3, PM2, PM5, PP5_STRONG	-	9708	-	unknown	Panel	negative	m	50.47
LHON 1219	*MT-ND1*	NC_012920.1	m.3460G>A	p.(Ala52Thr)	NC_012920.1:m.3460G>A (heteroplasmic, 98.0% of reads)	na	MT	P	PP3, PM2, PP5_STRONG	-	9722	-	unknown	no	unknown	m	51.94
LHON 1221	*MT-ND4*	NC_012920.1	m.11778G>A	p.(Arg340His)	NC_012920.1:m.11778G>A (heteroplasmic, 98.5% of reads)	na	MT	P	PP3, PM2, PM5, PP5_STRONG	-	9708	-	unknown	no	negative	m	50.61
LHON 1221	*OPA1*	NM_015560.3	c.*322G>A	p.(?)	NC_000003.11:g.193412711G>A	het	AR+AD	VUS	PM2, BP7	-	-	-	unknown	no	negative	m	50.61
MDS 138	*RP11*	NM_178857.5	c.133C>T	p.(Arg45Trp)	NC_000008.10:g.10480579G>A	het	AR+AD	LP	PM2, PP5_STRONG	CM105618	2193	0.0001	unknown	no	unknown	f	49.12
MDS 138	*RP11*	NM_178857.5	c.449C>T	p.(Thr150Ile)	NC_000008.10:g.10480263G>A	het	AR+AD	VUS	PM2	CM131357	-	0.0001	unknown	no	unknown	f	49.12
MDS 163	*GUCA1A*	NM_000409.3	c.359G>T	p.(Arg120Leu)	NC_000006.11:g.42146547G>T	het	AD	LP	PM2, PM1_SUPPORTING, PP2, PP5_STRONG	CM2036694	802212	-	unknown	Panel	positive (triplex)	f	38.31
MDS 166	*RP11*	NM_178857.5	c.133C>T	p.(Arg45Trp)	NC_000008.10:g.10480579G>A	het	AR+AD	LP	PM2, PP5_STRONG	CM105618	2193	0.0001	unknown	Panel	positive (triplex)	m	51.89
MDS 241	*RP11*	NM_178857.5	c.133C>T	p.(Arg45Trp)	NC_000008.10:g.10480579G>A	het	AR+AD	LP	PM2, PP5_STRONG	CM105618	2193	0.0001	unknown	no	negative	f	55.44
MDS 290	*BEST1*	NM_004183.3	c.779del	p.(Pro260Glnfs*29)	NC_000011.9:g.61725682del	hom	AR+AD	P	PM2, PVS1, PP5	CD033244	817618	0.0001	unknown	Panel	positive (duplex)	f	43.49
MDS 360	*ABCA4*	NM_000350.2	c.2829del	p.(Pro944Glnfs*6)	NC_000001.10:g.94512565del	het	AR	P	PM2, PVS1, PP5	CD1310357	-	-	unknown	no	negative	m	58.19
MDS 360	*ABCA4*	NM_000350.2	c.5351T>G	p.(Leu1784Arg)	NC_000001.10:g.94480208A>C	het	AR	LP	PP3, PM2, PM1, PP2, PP5	CM2011138	-	0.0001	unknown	no	negative	m	58.19
MDS 383	*ABCA4*	NM_000350.2	c.1937+1G>A	p.(?)	NC_000001.10:g.94528132C>T	het	AR	P	PM2, PVS1_STRONG, PP5_STRONG	CS003487	-	0.0001	unknown	Panel	negative	m	42.49
MDS 383	*ABCA4*	NM_000350.2	c.5603A>T	p.(Asn1868Ile)	NC_000001.10:g.94476467T>A	het	AR	VUS	PP2, PP5, BA1, BS2	CM015091	-	0.0426	unknown	Panel	negative	m	42.49
MDS 383	*PRPF8*	NM_006445.3	c.1898G>A	p.(Gly633Asp)	NC_000017.10:g.1580945C>T	het	AD	VUS	PM2, PP2	-	-	-	unknown	Panel	negative	m	42.49
MDS 391	*ABCA4*	NM_000350.2	c.3287C>T	p.(Ser1096Leu)	NC_000001.10:g.94508358G>A	hom	AR	P	PP3, PM2, PM1, PP2, PP5_STRONG	CM1311147	1184970	0.0001	unknown	no	negative	m	58.03
MDS 396	*ABCA4*	NM_000350.2	c.5461-10T>C	p.(?)	NC_000001.10:g.94476951A>G	het	AR	P	PM2, PP5_STRONG, BP4, PS4_MODERATE	CS057513	1048134	0.0002	paternal	no	negative	f	53.83
MDS 396	*ABCA4*	NM_000350.2	c.5603A>T	p.(Asn1868Ile)	NC_000001.10:g.94476467T>A	het	AR	VUS	PP2, PP5, BA1, BS2	CM015091	1048134	0.0426	unknown	no	negative	f	53.83
MDS 396	*ABCA4*	NM_000350.2	c.5882G>A	p.(Gly1961Glu)	NC_000001.10:g.94473807C>T	het	AR	P	PP3, PM2, PM5, PP2, PP5_STRONG, PS4_MODERATE	CM970016	635154	0.0047	maternal	no	negative	f	53.83
MDS 397	*intergenic*	NC_000006.11	g.100040987G>C	na	NC_000006.11:g.100040987G>C	het	AD	LP	PM2, PP5_STRONG, BP7	CR160157	-	0.000007	unknown	no	positive (duplex)	m	54.06
MDS 401	*GUCA1A*	NM_000409.3	c.265G>A	p.(Glu89Lys)	NC_000006.11:g.42146081G>A	het	AD	LP	PM2, PM1, PP2, PP5	CM094672	986883	-	unknown	no	negative	f	58.07
MDS 402	*PRPH2*	NM_000322.4	c.728T>C	p.(Leu243Pro)	NC_000006.11:g.42672203A>G	het	AD	LP	PP3, PM2, PM1, PP2	-	-	-	unknown	no	negative	m	57.17
MDS 404	*PRPH2*	NM_000322.4	c.514C>T	p.(Arg172Trp)	NC_000006.11:g.42689559G>A	het	AD	P	PM2, PM5, PP2, PP5_STRONG	CM930639	-	-	unknown	no	negative	f	55.76
MDS 404	*ROM1*	NM_000327.3	c.685C>T	p.(Arg229Cys)	NC_000011.9:g.62381824C>T	het	AD	VUS	PM2, BP4	-	-	0.000004	unknown	no	negative	f	55.76
MDS 407	*ABCA4*	NM_000350.2	c.5882G>A	p.(Gly1961Glu)	NC_000001.10:g.94473807C>T	het	AR	P	PP3, PM2, PM5, PP2, PP5_STRONG, PS4_MODERATE	CM970016	635154	0.0047	maternal	no	negative	f	54.38
MDS 407	*ABCA4*	NM_000350.2	c.6426_6428del	p.(Ile2142del)	NC_000001.10:g.94466446_94466448del	het	AR	LP	PM2, PM4, PM1	-	-	-	Father negative for variant	no	negative	f	54.38
MDS 407	*ABCA4*	NM_000350.2	c.3758C>T	p.(Thr1253Met)	NC_000001.10:g.94502756G>A	het	AR	VUS	PM2, PP2, BP6	CM012036	-	0.0004	maternal	no	negative	f	54.38
MDS 408	*PRPH2*	NM_000322.4	c.515G>A	p.(Arg172Gln)	NC_000006.11:g.42689558C>T	het	AD	P	PS1_MODERATE, PM2, PM5, PP2, PP5_STRONG	CM930637	13167	0.000004	unknown	no	negative	f	61.14
MDS 408	*ABCA4*	NM_000350.2	c.5603A>T	p.(Asn1868Ile)	NC_000001.10:g.94476467T>A	het	AR	VUS	PP2, PP5, BA1, BS2	CM015091	-	0.0426	unknown	no	negative	f	61.14
MDS 408	*ABCA4*	NM_000350.2	c.1267_1268inv	p.(His423Cys)	NC_000001.10:g.94544234_94544235inv	het	AR	VUS	PM2, PP2	-	-	-	unknown	no	negative	f	61.14
MDS 411	*MT-TL1*	NC_012920.1	m.3243A>G	-	NC_012920.1:m.3243A>G (heteroplasmic, 18.7% of reads)	na	MT	P	PP3, PM2, PM5, PP5_STRONG	-	9589	-	unknown	no	negative	f	47.07
MDS 411	*ABCA4*	NM_000350.2	c.5714+5G>A	p.(?)	NC_000001.10:g.94476351C>T	het	AR	P	PP3, PM2, PP5_STRONG, PS4_MODERATE	CS982057	-	0.0003	unknown	no	negative	f	47.07
MDS 413	*ABCA4*	NM_000350.2	c.1957C>T	p.(Arg653Cys)	NC_000001.10:g.94526296G>A	het	AR	P	PP3, PM2, PM5, PM1, PP2, PP5_STRONG	CM003369	-	0.000016	unknown	no	positive (duplex)	m	49.98
MDS 413	*ABCA4*	NM_000350.2	c.4253+43G>A	p.(?)	NC_000001.10:g.94496509C>T	het	AR	LP	PM2, PP5_STRONG, BP7	CS193151	-	0.0047	unknown	no	positive (duplex)	m	49.98
MDS 414	*CDHR1*	NM_033100.3	c.783G>A	p.(Pro261=); p.(Asp214_Pro261del)	NC_000010.10:g.85962879G>A	hom	AR	LP	PP3, PM2, PP5_STRONG	CM188677&CS140565	301224	0.0045	unknown	no	negative	m	50.91
MDS 417	*ABCA4*	NM_000350.2	c.1622T>C	p.(Leu541Pro)	NC_000001.10:g.94528806A>G	het	AR	P	PP3, PM2, PP2, PP5_STRONG, PS4_MODERATE	CM990022	7901	0.0003	unknown	no	negative	m	53.08
MDS 417	*ABCA4*	NM_000350.2	c.3113C>T	p.(Ala1038Val)	NC_000001.10:g.94508969G>A	het	AR	P	PM2, PM1_SUPPORTING, PP2, PP5_STRONG, PS4_MODERATE	CM970006	7901	0.0024	unknown	no	negative	m	53.08
MDS 417	*ABCA4*	NM_000350.2	c.5882G>A	p.(Gly1961Glu)	NC_000001.10:g.94473807C>T	het	AR	P	PP3, PM2, PM5, PP2, PP5_STRONG, PS4_MODERATE	CM970016	635154	0.0047	unknown	no	negative	m	53.08
MDS 418	*KLHL7*	NM_001031710.2	c.969del	p.(Phe323Leufs*15)	NC_000007.13:g.23205349del	het	AD	LP	PM2, PVS1	-	-	-	unknown	no	negative	f	54.88
MDS 420	*RP11*	NM_178857.5	c.133C>T	p.(Arg45Trp)	NC_000008.10:g.10480579G>A	het	AD+AR	LP	PM2, PP5_STRONG	CM105618	2193	0.0001	unknown	no	negative	m	53.82
MDS 420	*RP11*	NM_178857.5	c.2873G>A	p.(Arg958His)	NC_000008.10:g.10468735C>T	het	AD+AR	VUS	PM2, BS2_SUPPORTING	CM131343	-	0.00002	unknown	no	negative	m	53.82
MDS 420	*RP11*	NM_178857.5	c.449C>T	p.(Thr150Ile)	NC_000008.10:g.10480263G>A	het	AD+AR	VUS	PM2	CM131357	-	0.0001	unknown	no	negative	m	53.82
MDS 426	*ABCA4*	NM_000350.2	c.45G>A	p.(Trp15*)	NC_000001.10:g.94586557C>T	het	AR	P	PM2, PVS1, PP5	CM990010	-	0.0001	unknown	no	negative	f	44.04
MDS 426	*ABCA4*	NM_000350.2	c.5603A>T	p.(Asn1868Ile)	NC_000001.10:g.94476467T>A	het	AR	VUS	PP2, PP5, BA1, BS2	CM015091	-	0.0426	unknown	no	negative	f	44.04
MDS 427	*CRX*	NM_000554.4	na	na	NC_000019.9:g.48340644_48406926del	het	AD	LP	PM2, PVS1	na	-	na	unknown	no	positive (duplex)	f	49.31
MDS 431	*ABCC6*	NM_001171.5	c.2996-1724_4209-479del	p.(?)	NC_000016.9:g.16245124_16261530del	hom	AR	LP	PM2, PVS1	na	-	na	unknown	no	negative	m	51.73
MDS 435	*CRB1*	NM_201253.2	c.498_506del	p.(Ile167_Gly169del)	NC_000001.10:g.197297979_197297987del	het	AR	P	PM2, PM4, PP5_STRONG, PS4_MODERATE	CD061397	-	0.0006	paternal	no	negative	m	48.34
MDS 435	*CRB1*	NM_201253.2	c.1712A>C	p.(Glu571Ala)	NC_000001.10:g.197390670A>C	het	AR	LP	PM2, PM1_SUPPORTING, PP2, PP5, PS4_MODERATE	CM173694	-	0.0001	maternal	no	negative	m	48.34
MDS 438	*POC1B*	NM_172240.2	c.1033-327T>A	p.(I345Afs*9)	NC_000012.11:g.89853822A>T	het	AR	LP	PM2, PS3, PM3, BP4	-	-	0.0001	unknown	no	negative	f	50.44
MDS 438	*POC1B*	NM_172240.2	c.810+1G>T	p.(?)	NC_000012.11:g.89864137C>A	het	AR	LP	PM2, PVS1	CS148091	-	0.0001	unknown	no	negative	f	50.44
MDS 439	*ABCA4*	NM_000350.2	c.4537dup	p.(Gln1513Profs*42)	NC_000001.10:g.94495009dup	het	AR	P	PM2, PVS1, PP5, PS4_MODERATE	CI014469	-	0.0001	unknown	no	negative	f	47.91
MDS 439	*ABCA4*	NM_000350.2	c.5882G>A	p.(Gly1961Glu)	NC_000001.10:g.94473807C>T	het	AR	P	PP3, PM2, PM5, PP2, PP5_STRONG, PS4_MODERATE	CM970016	635154	0.0047	unknown	no	negative	f	47.91
MDS 440	*CDHR1*	NM_033100.3	c.1381C>T	p.(Gln461*)	NC_000010.10:g.85970817C>T	het	AR	P	PM2, PVS1, PP5	CM129171	-	-	unknown	no	negative	f	48.28
MDS 440	*CDHR1*	NM_033100.3	c.783G>A	p.(Pro261=); p.(Asp214_Pro261del)	NC_000010.10:g.85962879G>A	het	AR	LP	PP3, PM2, PP5_STRONG	CM188677&CS140565	301224	0.0045	unknown	no	negative	f	48.28
MDS 441	*PRPH2*	NM_000322.4	c.514C>T	p.(Arg172Trp)	NC_000006.11:g.42689559G>A	het	AD	P	PM2, PM5, PP2, PP5_STRONG	CM930639	-	-	unknown	no	positive (duplex)	f	46.40
MDS 444	*PRPH2*	NM_000322.4	c.520T>G	p.(Trp174Gly)	NC_000006.11:g.42689553A>C	het	AD	LP	PP3, PM2, PM5_SUPPORTING, PM1, PP2	-	-	-	unknown	no	negative	f	49.16
MDS 444	*ABCC6*	NM_001171.5	c.2278C>T	p.(Arg760Trp)	NC_000016.9:g.16272792G>A	het	AR	P	PP3, PM2, PM5_SUPPORTING, PM1, PP2, PP5_STRONG	CM050523	-	0.0001	unknown	no	negative	f	49.16
MDS 448	*CDHR1*	NM_033100.3	c.783G>A	p.(Pro261=); p.(Asp214_Pro261del)	NC_000010.10:g.85962879G>A	hom	AR	LP	PP3, PM2, PP5_STRONG	CM188677&CS140565	301224	0.0045	unknown	no	positive (duplex)	f	53.29
MDS 448	*CRB1*	NM_201253.2	c.500T>G	p.(Ile167Ser)	NC_000001.10:g.197297981T>G	het	AR	VUS	PM2, PP2	-	-	-	unknown	no	positive (duplex)	f	53.29
MDS 449	*RP11*	NM_178857.5	c.133C>T	p.(Arg45Trp)	NC_000008.10:g.10480579G>A	het	AD+AR	LP	PM2, PP5_STRONG	CM105618	2193	0.0001	unknown	no	unknown	f	56.09
MDS 450-1	*intergenic*	NC_000006.11	g.100040987G>C	na	NC_000006.11:g.100040987G>C	het	AD	LP	PM2, PP5_STRONG, BP7	CR160157	-	0.000007	unknown	no	positive (duplex)	f	56.32
MDS 450-2	*intergenic*	NC_000006.11	g.100040987G>C	na	NC_000006.11:g.100040987G>C	het	AD	LP	PM2, PP5_STRONG, BP7	CR160157	-	0.000007	unknown	no	positive (duplex)	f	54.60
MDS 452	*CDHR1*	NM_033100.3	c.783G>A	p.(Pro261=); p.(Asp214_Pro261del)	NC_000010.10:g.85962879G>A	hom	AR	LP	PP3, PM2, PP5_STRONG	CM188677&CS140565	301224	0.0045	unknown	single genes	positive (duplex)	m	53.14
MDS 453	*IMPG2*	NM_016247.3	c.68dup	p.(Asp23Glufs*29)	NC_000003.11:g.101039149dup	het	AD	P	PM2, PVS1, PP5	CI1613913	236460	0.0001	unknown	no	negative	f	55.71
MDS 457	*ABCA4*	NM_000350.2	c.1622T>C	p.(Leu541Pro)	NC_000001.10:g.94528806A>G	het	AR	P	PP3, PM2, PP2, PP5_STRONG, PS4_MODERATE	CM990022	7901	0.0003	unknown	no	unknown	f	66.32
MDS 457	*ABCA4*	NM_000350.2	c.3113C>T	p.(Ala1038Val)	NC_000001.10:g.94508969G>A	het	AR	P	PM2, PM1_SUPPORTING, PP2, PP5_STRONG, PS4_MODERATE	CM970006	7901	0.0024	unknown	no	unknown	f	66.32
MDS 457	*ABCA4*	NM_000350.2	c.5603A>T	p.(Asn1868Ile)	NC_000001.10:g.94476467T>A	het	AR	VUS	PP2, PP5, BA1, BS2	CM015091	-	0.0426	unknown	no	unknown	f	66.32
MDS 459	*PRPH2*	NM_000322.4	c.934del	p.(Val312Cysfs*12)	NC_000006.11:g.42666140del	het	AD	P	PM2, PVS1_STRONG, PP5_STRONG	-	865787	-	unknown	no	positive (multiplex)	f	35.19
MDS 464	*ABCA4*	NM_000350.2	c.5882G>A	p.(Gly1961Glu)	NC_000001.10:g.94473807C>T	het	AR	P	PP3, PM2, PM5, PP2, PP5_STRONG, PS4_MODERATE	CM970016	635154	0.0047	unknown	no	negative	m	38.3
MDS 464	*ABCA4*	NM_000350.2	c.6088C>T	p.(Arg2030*)	NC_000001.10:g.94471056G>A	het	AR	P	PM2, PVS1, PP5, PS4	CM990071	-	0.00003	unknown	no	negative	m	38.3
MDS 466	*CRX*	NM_000554.4	c.766C>T	p.(Gln256*)	NC_000019.9:g.48343090C>T	het	AD	P	PM2, PVS1, PP5, PS4	CM1514251	-	-	unknown	no	negative	f	40.35
MDS 467	*RPGR*	NM_001034853.1	c.3373C>T	p.(Gln1125*)	NC_000023.10:g.38144879G>A	hemi	XLD+XLR	P	PM2, PVS1, PP5, PS4	CM2036779	-	-	unknown	no	negative	m	35.91
MDS 468	*CLN3*	NM_001042432.1	c.461-280_677+382del	p.(?)	NC_000016.9:g.28497286_28498251del	hom	AR	LP	PM2, PVS1	na	-	na	unknown	no	negative	f	34.84
MISC 136	*GRK1*	NM_002929.2	na	na	NC_000013.10:g.114317313_114325241del	hom	AR	LP	PM2, PVS1	na	-	na	unknown	no	unknown	f	50.19
MISC 219	*MT-ATP6*	NC_012920.1	m.9032T>C	p.(Leu169Pro)	NC_012920.1:m.9032T>C (heteroplasmic, 34.0% of reads)	na	MT	LP	PM2, PP5_STRONG, PS4_MODERATE	-	-	-	unknown	Panel	positive (multiplex)	m	38.35
MISC 243	*ABCA4*	NM_000350.2	c.1532G>A	p.(Arg511His)	NC_000001.10:g.94543268C>T	het	AR	LP	PS1_MODERATE, PM5, PP2, PP5, BP6, PS4_MODERATE	CM1210006	-	0.0007	unknown	no	negative	m	54.43
MISC 243	*ABCA4*	NM_000350.2	c.2588G>C	p.(Gly863Ala)	NC_000001.10:g.94517254C>G	het	AR	P	PP3, PM2, PP2, PP5_STRONG, PS4_MODERATE	CM970003&CS024003	-	0.0044	unknown	no	negative	m	54.43
MISC 243	*ABCA4*	NM_000350.2	c.5603A>T	p.(Asn1868Ile)	NC_000001.10:g.94476467T>A	het	AR	VUS	PP2, PP5, BA1, BS2	CM015091	-	0.0426	unknown	no	negative	m	54.43
MISC 245	*NPHP1*	NM_207181.2	na, deletion whole NPHP1 gene	na	NC_000002.11, na	hom	AR	LP	PM2, PVS1	na	-	na	unknown	no	positive (duplex)	f	50.31
MISC 249	*MT-TL1*	NC_012920.1	m.3243A>G	-	NC_012920.1:m.3243A>G (heteroplasmic, 17.0% of reads)	na	MT	P	PP3, PM2, PM5, PP5_STRONG	-	-	-	unknown	Panel	positive (multiplex)	f	40.98
MISC 249	*BEST1*	NM_004183.3	c.584C>T	p.(Ala195Val)	NC_000011.9:g.61724418C>T	het	AD+AR	P	PP3, PM2, PP2, PP5_STRONG	CM004430	-	0.00025	unknown	Panel	positive (multiplex)	f	40.98
MISC 252-1	*IKBKG*	NM_001099857.1	na, deletion exon 4-10	na	NC_000023.10, na	het	XLD+XLR	LP	PM2, PVS1	na	-	na	maternal	no	positive (duplex)	f	51.41
MISC 252-2	*IKBKG*	NM_001099857.1	na, deletion exon 4-10	na	NC_000023.10, na	het	XLD+XLR	LP	PM2, PVS1	na	-	na	maternal	no	positive (duplex)	f	54.21
MISC 252-3	*IKBKG*	NM_001099857.1	na, deletion exon 4-10	na	NC_000023.10, na	het	XLD+XLR	LP	PM2, PVS1	na	-	na	unknown	no	positive (duplex)	f	51.13
MISC 262	*KCNV2*	NM_133497.3	na	na	NC_000009.11:g.2719299_2730682del	hom	AR	LP	PM2, PVS1	na	-	na	unknown	no	positive (duplex)	f	53.06
MISC 264	*PRPH2*	NM_000322.4	c.515G>C	p.(Arg172Pro)	NC_000006.11:g.42689558C>G	het	AD	P	PS1_MODERATE, PM2, PM5, PP2, PP5_STRONG	-	-	-	unknown	no	negative	f	52.45
MISC 265	*CYP4V2*	NM_207352.3	c.1199G>A	p.(Arg400His)	NC_000004.11:g.187130127G>A	hom	AR	P	PP3, PM2, PM5, PM1_SUPPORTING, PP2, PP5_STRONG	CM056577	39254	0.0001	unknown	no	positive (duplex)	m	50.92
MISC 265	*ABCA4*	NM_000350.2	c.769-784C>T	p.(?)	NC_000001.10:g.94549781G>A	het	AR	VUS	BP7	CS1914677	-	0.0028	unknown	no	positive (duplex)	m	50.92
MISC 267	*FZD4*	NM_012193.3	c.1474G>C	p.(Gly492Arg)	NC_000011.9:g.86662324C>G	het	AD	LP	PP3, PM2, PP5_STRONG	CM082724	-	-	unknown	no	positive (multiplex)	f	53.77
MISC 267	*CAPN5*	NM_004055.4	c.391T>A	p.(Trp131Arg)	NC_000011.9:g.76823728T>A	het	AD	VUS	PP3, PM2	-	-	0.000004	unknown	no	positive (multiplex)	f	53.77
MISC 269	*GPR143*	NM_000273.2	c.232G>A	p.(Asp78Asn)	NC_000023.10:g.9733626C>T	het	XLR+XLD	P	PP3, PM2, PM5_SUPPORTING, PM1_SUPPORTING, PP2, PP5_STRONG	CM003027	-	-	unknown	no	unknown	f	49.44
MISC 269	*RPGRIP1*	NM_020366.3	c.800+1G>A	p.(?)	NC_000014.8:g.21771703G>A	het	AR	P	PM2, PVS1_STRONG, PP5_STRONG	CS057789	-	0.0001	unknown	no	unknown	f	49.44
MISC 272-1	*MFSD8*	NM_152778.2	c.750A>G	p.(Arg233Serfs*5)	NC_000004.11:g.128859942T>C	hom	AR	P	PM2, PS3, PP1_STRONG	-	-	-	unknown	no	positive (duplex)	f	48.53
MISC 272-2	*MFSD8*	NM_152778.2	c.750A>G	p.(Arg233Serfs*5)	NC_000004.11:g.128859942T>C	hom	AR	P	PM2, PS3, PP1_STRONG	-	-	-	unknown	no	positive (duplex)	m	40.33
MISC 293	*RNU4ATAC*	NC_000002.11	g.122288468C>T	na	NC_000002.11:g.122288468C>T	het	AR	P	PM2, PP5_STRONG, PS4_MODERATE	CR1512533	218083	0.0005	unknown	Panel	unknown	m	52.57
MISC 293	*RNU4ATAC*	NC_000002.11	g.122288503G>A	na	NC_000002.11:g.122288503G>A	het	AR	LP	PM2, PP5_STRONG	CR1512535	-	0.0001	unknown	Panel	unknown	m	52.57
MISC 295	*FZD4*	NM_012193.3	c.1501_1502del	p.(Leu501Serfs*33)	NC_000011.9:g.86662298_86662299del	het	AD	P	PM2, PVS1_STRONG, PP5_STRONG	CD022549	-	-	unknown	no	unknown	m	44.61
MISC 296	*COL18A1*	NM_030582.3	c.4054_4055del	p.(Leu1352Valfs*72)	NC_000021.8:g.46930005_46930006del	hom	AR	P	PM2, PVS1_STRONG, PP5_STRONG	CD023246	65410	0.0003	unknown	no	negative	m	51.45
MISC 303	*PHYH*	NM_006214.3	c.529G>A	p.(Asp177Asn)	NC_000010.10:g.13330509C>T	hom	AR	P	PP3, PM2, PM5, PM1, PP5_STRONG	-	-	-	paternal/unknown	no	positive (duplex)	m	37.50
MISC 304	*MMP19*	NM_002429.5	na	na	NC_000012.11:g.56238545_56245997[3]	het	AD	LP	PM2, PVS1	CP152029	-	na	unknown	no	positive (multiplex)	m	55.81
MISC 305	*HADHA*	NM_000182.4	c.1528G>C	p.(Glu510Gln)	NC_000002.11:g.26418053C>G	hom	AR	P	PP3, PM2, PP5_STRONG	CM940884	100085	0.0018	unknown	no	negative	f	56.91
MISC 306	*ARID1B*	NM_020732.3	c.4221del	p.(Tyr1407*)	NC_000006.11:g.157521949del	hom	AD	P	PM2, PVS1, PP5	-	1275782	-	unknown	single genes	negative	f	42.46
MISC 313	*CNGB3*	NM_019098.4	c.1148del	p.(Thr383Ilefs*13)	NC_000008.10:g.87656009del	het	AR	P	PM2, PVS1, PP5, PS4_MODERATE	CD001927	-	0.0020	unknown	no	positive (triplex)	f	58.29
MISC 313	*CNGB3*	NM_019098.4	c.1208G>A	p.(Arg403Gln)	NC_000008.10:g.87645092C>T	het	AR	VUS	PP3, PP5, BS1, BS2, BP6	CM043918	-	0.0046	unknown	no	positive (triplex)	f	58.29
MISC 313	*PAX3*	NM_181458.3	c.1097del	p.(Tyr366Leufs*15)	NC_000002.11:g.223084935del	het	AD	P	PM2, PVS1, PP5	-	1275818	-	unknown	no	positive (triplex)	f	58.29
MISC 314	*MT-ATP6*	NC_012920.1	m.8993T>G	p.(Leu156Arg)	NC_012920.1:m.8993T>G (heteroplasmic, 57.9% of reads)	na	MT	P	PP3, PM5, PP5_STRONG	-	-	-	unknown	no	positive (duplex)	f	39.51
MISC 314	*ABCC6*	NM_001171.5	c.1501A>T	p.(Ile501Phe)	NC_000016.9:g.16284155T>A	het	AR	VUS	PM2, PP2	-	-	0.0001	unknown	no	positive (duplex)	f	39.51
MISC 314	*IMPG2*	NM_016247.3	c.3258G>T	p.(Trp1086Cys)	NC_000003.11:g.100949965C>A	het	AD	VUS	PM2	-	-	-	unknown	no	positive (duplex)	f	39.51
MISC 315	*TUBB4B*	NM_006088.5	c.1172G>A	p.(Arg391His)	NC_000009.11:g.140137842G>A	het	AD	P	PP3, PM2, PM5, PP2, PP5_STRONG	CM1716631	492938	-	unknown	single genes	negative	m	32.61
MISC 315	*MYO15A*	NM_016239.3	c.1185dup	p.(Glu396Argfs*36)	NC_000017.10:g.18023299dup	het	AR	P	PM2, PVS1, PP5	CI121691	-	0.0001	unknown	single genes	negative	m	32.61
MISC 320	*NR2E3*	NM_014249.3	c.932G>A	p.(Arg311Gln)	NC_000015.9:g.72105913G>A	het	AR+AD	P	PP3, PM2, PM5, PP2, PP5_STRONG	CM000538	-	0.0004	unknown	no	negative	m	36.99
MISC 320	*NR2E3*	NM_014249.3	c.248G>A	p.(Cys83Tyr)	NC_000015.9:g.72104108G>A	het	AR+AD	P	PP3, PM2, PM5, PP2, PP5_STRONG	CM105189	-	0.00001	unknown	no	negative	m	36.99
MISC 320	*RP11*	NM_178857.5	na	na	NC_000008.10:g.10445159_10466425del	het	AR+AD	LP	PM2, PVS1	na	-	na	unknown	no	negative	m	36.99
NYS 19	*FRMD7*	NM_194277.2	c.781C>T	p.(Arg261*)	NC_000023.10:g.131216515G>A	het	XLR+XLD	P	PVS1_STRONG, PM2_MODERATE, PP5_SUPPORTING	-	-	-	unknown	no	negative	f	47.77
OA 16	*TYR*	NM_000372.4	c.1205G>A	p.(Arg402Gln)	NC_000011.9:g.89017961G>A	het	AR	VUS	PM5_SUPPORTING, PP2, PP5, BP6	CM971555	-	0.1769	unknown	no	positive (duplex)	f	45.82
OA 16	*TYR*	NM_000372.4	c.325G>A	p.(Gly109Arg)	NC_000011.9:g.88911446G>A	het	AR	P	PP3, PM2, PM1, PP2, PP5_STRONG	CM013052	-	0.00008	unknown	no	positive (duplex)	f	45.82
OA 17-1	*TYR*	NM_000372.4	c.902C>G	p.(Pro301Arg)	NC_000011.9:g.88924452C>G	het	AR	LP	PP3, PM2, PM5_SUPPORTING, PM1, PP2	-	-	-	unknown	no	positive (duplex)	f	46.25
OA 17-1	*TYR*	NM_000372.4	c.1205G>A	p.(Arg402Gln)	NC_000011.9:g.89017961G>A	het	AR	VUS	PM5_SUPPORTING, PP2, PP5, BP6	CM971555	-	0.1769	unknown	no	positive (duplex)	f	46.25
OA 17-2	*TYR*	NM_000372.4	c.902C>G	p.(Pro301Arg)	NC_000011.9:g.88924452C>G	het	AR	LP	PP3, PM2, PM5_SUPPORTING, PM1, PP2	-	-	-	unknown	no	positive (duplex)	f	46.24
OA 17-2	*TYR*	NM_000372.4	c.1205G>A	p.(Arg402Gln)	NC_000011.9:g.89017961G>A	het	AR	VUS	PM5_SUPPORTING, PP2, PP5, BP6	CM971555	-	0.1769	unknown	no	positive (duplex)	f	46.24
OA 18	*TYR*	NM_000372.4	c.1205G>A	p.(Arg402Gln)	NC_000011.9:g.89017961G>A	het	AR	VUS	PM5_SUPPORTING, PP2, PP5, BP6	CM971555	-	0.1769	maternal	no	negative	f

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SRP 1124	*RP11*	NM_178857.5	c.4150G>T	p.(Gly1384*)	NC_000008.10:g.10467458C>A	het	AD+AR	LP	PM2, PVS1_STRONG	-	-	-	unknown	no	negative	m	57.52
SRP 1124	*RP11*	NM_178857.5	c.4354G>A	p.(Glu1452Lys)	NC_000008.10:g.10467254C>T	het	AD+AR	VUS	PM2, BP4	-	-	-	unknown	no	negative	m	57.52
SRP 1125	***EYS***	**NM_001142800.1**	**c.6714del**	**p.(Ile2239Serfs*17)**	**NC_000006.11:g.64776242del**	**het**	**AR**	**P**	**PM2, PVS1, PP5_STRONG**	**CD088088**	**357699**	**0.0001**	**unknown**	**no**	**negative**	**f**	**32.81**
SRP 1125	***EYS***	**NM_001142800.1**	**c.8984T>A**	**p.(Ile2995Asn)**	**NC_000006.11:g.64430943A>T**	**het**	**AR**	**LP**	**PM2, PP5_STRONG**	**CM103600**	**939428**	**0.000006**	**unknown**	**no**	**negative**	**f**	**32.81**
SRP 1125	*7q31.31q31.3*	na	na	na	NC_000007.13:g.119629529_122059846del	het	AD	LP	PM2, PVS1	na	-	na	unknown	no	negative	f	32.81
SRP 1127	***EYS***	**NM_001142800.1**	**c.8984T>A**	**p.(Ile2995Asn)**	**NC_000006.11:g.64430943A>T**	**hom**	**AR**	**LP**	**PM2, PP5_STRONG**	**CM103600**	**939428**	**0.000006**	**unknown**	**no**	**negative**	**f**	**49.80**
SRP 1128	***CDHR1***	**NM_033100.3**	**c.2522_2528del**	**p.(Ile841Serfs*119)**	**NC_000010.10:g.85974319_85974325del**	**hom**	**AR**	**P**	**PM2, PVS1_MODERATE, PP5_STRONG, PS4_MODERATE**	**CD160506**	**194793**	**0.0001**	**unknown**	**no**	**negative**	**m**	**57.35**
SRP 1128	*RGR*	NM_002921.3	c.196A>C	p.(Ser66Arg)	NC_000010.10:g.86007463A>C	hom	AR+AD	VUS	PM2	CM993198	-	0.0001	unknown	no	negative	m	57.35
SRP 1130	***PRPF4***	**NM_004697.4**	**c.944C>T**	**p.(Pro315Leu)**	**NC_000009.11:g.116050463C>T**	**het**	**AD**	**LP**	**PM2, PP5_STRONG**	**CM144902**	**143057**	**0.000008**	**unknown**	**no**	**negative**	**m**	**56.00**
SRP 1130	*RPE65*	NM_000329.2	c.10C>T	p.(Gln4*)	NC_000001.10:g.68915579G>A	het	AR	LP	PM2, PVS1	-	-	-	unknown	no	negative	m	56.00
SRP 1130	*RPE65*	NM_000329.2	c.409T>C	p.(Tyr137His)	NC_000001.10:g.68910300A>G	het	AR	VUS	PP3, PM2, PP2	-	-	-	unknown	no	negative	m	56.00
SRP 1131	***EYS***	**NM_001142800.1**	**c.3444-31214_3685-8848dup**	**p.(?)**	**NC_000006.11:g.65312052_65367354dup**	**het**	**AR**	**LP**	**PM2, PVS1**	**na**	**-**	**na**	**paternal**	**no**	**negative**	**f**	**54.47**
SRP 1131	***EYS***	**NM_001142800.1**	**na**	**na**	**NC_000006.11:g.65647741_65658294delins[65653992_65654056inv;CAATTTTGTAT]**	**het**	**AR**	**LP**	**PM2, PVS1**	**na**	**-**	**na**	**maternal**	**no**	**negative**	**f**	**54.47**
SRP 1134	***USH2A***	**NM_206933.2**	**c.2276G>T**	**p.(Cys759Phe)**	**NC_000001.10:g.216420460C>A**	**het**	**AR**	**P**	**PP3, PM2, PM1_SUPPORTING, PP5_STRONG, PS4_MODERATE**	**CM001372**	**2356**	**0.0010**	**unknown**	**no**	**negative**	**f**	**49.22**
SRP 1134	***USH2A***	**NM_206933.2**	**c.2299del**	**p.(Glu767Serfs*21)**	**NC_000001.10:g.216420437del**	**het**	**AR**	**P**	**PM2, PVS1, PP5, PS4**	**CD982997**	**560516**	**0.0008**	**unknown**	**no**	**negative**	**f**	**49.22**
SRP 1134	*USH2A*	NM_206933.2	c.4714C>T	p.(Leu1572Phe)	NC_000001.10:g.216270469G>A	het	AR	VUS	PP3, PM2, BP6_STRONG	CM119884	-	0.0008	unknown	no	negative	f	49.22
SRP 1139	***PRPF31***	**NM_015629.3**	**na**	**na**	**NC_000019.9:g.54602638_54636555del**	het	AD	**LP**	**PM2, PVS1**	**na**	**na**	**na**	unknown	**Panel**	**positive (duplex)**	**f**	**53.80**
SRP 1145	***CRB1***	**NM_201253.2**	**c.2914G>T**	**p.(Glu972*)**	**NC_000001.10:g.197403907G>T**	**het**	**AR**	**P**	**PM2, PVS1, PP5**	**-**	**-**	**-**	**unknown**	**no**	**negative**	**m**	**46.82**
SRP 1145	***CRB1***	**NM_201253.2**	**c.498_506del**	**p.(Ile167_Gly169del)**	**NC_000001.10:g.197297979_197297987del**	**het**	**AR**	**P**	**PM2, PM4, PP5_STRONG, PS4_MODERATE**	**CD061397**	**96659**	**0.0006**	**unknown**	**no**	**negative**	**m**	**46.82**
SRP 1146	***TULP1***	**NM_003322.3**	**c.1024C>T**	**p.(Arg342*)**	**NC_000006.11:g.35473606G>A**	**het**	**AR**	**P**	**PM2, PVS1, PP5**	**-**	**-**	**-**	**unknown**	**no**	**negative**	**f**	**51.08**
SRP 1146	***TULP1***	**NM_003322.3**	**c.1496-6C>A**	**p.(?)**	**NC_000006.11:g.35466243G>T**	**het**	**AR**	**LP**	**PM2, PP5_STRONG, BP4, PS4_MODERATE**	**CS984713**	**-**	**0.0001**	**unknown**	**no**	**negative**	**f**	**51.08**
SRP 1147	***RP1***	**NM_006269.1**	**c.2029C>T**	**p.(Arg677*)**	**NC_000008.10:g.55538471C>T**	**het**	**AD+AR**	**P**	**PM2, PVS1_STRONG, PP5_STRONG, PS4_MODERATE**	**CM991103**	**-**	**-**	**unknown**	**no**	**negative**	**f**	**53.13**
SRP 1149	***EYS***	**NM_001142800.1**	**c.7723+1G>A**	**p.(?)**	**NC_000006.11:g.64497997C>T**	**het**	**AR**	**P**	**PM2, PVS1, PP5**	**CS172990**	**986903**	**-**	**unknown**	**no**	**negative**	**f**	**52.16**
SRP 1149	***EYS***	**NM_001142800.1**	**c.8648_8649del**	**p.(Thr2883Metfs*6)**	**NC_000006.11:g.64431281_64431282del**	**het**	**AR**	**LP**	**PM2, PVS1**	**-**	**-**	**-**	**unknown**	**no**	**negative**	**f**	**52.16**
SRP 1150	***NPHP1***	**NM_207181.2**	**na, deletion whole NPHP1 gene**	**na**	**NC_000002.11, na**	**hom**	**AR**	**LP**	**PM2, PVS1**	**na**	**636006**	**na**	**unknown**	**no**	**negative**	**f**	**46.02**
SRP 1151	***CEP290***	**NM_025114.3**	**c.5587-1G>C**	**p.(?)**	**NC_000012.11:g.88471122C>G**	**hom**	**AR**	**P**	**PM2, PVS1_STRONG, PP5_STRONG**	**CS072147**	**-**	**0.0001**	**unknown**	**no**	**negative**	**f**	**54.87**
SRP 1152	***ABCA4***	**NM_000350.2**	**c.4469G>A**	**p.(Cys1490Tyr)**	**NC_000001.10:g.94495071C>T**	**het**	**AR**	**P**	**PP3, PM2, PM5, PP2, PP5_STRONG, PS4_MODERATE**	**CM990056**	**99288**	**0.0001**	**unknown**	**no**	**negative**	**f**	**47.87**
SRP 1152	***ABCA4***	**NM_000350.2**	**c.5461-10T>C**	**p.(?)**	**NC_000001.10:g.94476951A>G**	**het**	**AR**	**P**	**PM2, PP5_STRONG, BP4, PS4_MODERATE**	**CS057513**	**1048134**	**0.0002**	**unknown**	**no**	**negative**	**f**	**47.87**
SRP 1152	*ABCA4*	NM_000350.2	c.5603A>T	p.(Asn1868Ile)	NC_000001.10:g.94476467T>A	hom	AR	VUS	PP2, PP5, BA1, BS2	CM015091	-	0.0426	unknown	no	negative	f	47.87
SRP 1153	***C1QTNF5***	**NM_001278431.1**	**c.489C>A**	**p.(Ser163Arg)**	**NC_000011.9:g.119210284G>T**	**het**	**AD**	**P**	**PS1, PM2, PP5_STRONG**	**CM1713958**	**-**	**-**	**unknown**	**no**	**negative**	**f**	**48.31**
SRP 1155	***CRB1***	**NM_201253.2**	**c.2290C>T**	**p.(Arg764Cys)**	**NC_000001.10:g.197396745C>T**	**het**	**AR**	**P**	**PM2, PM5, PM1, PP2, PP5_STRONG**	**CM992151**	**5732**	**0.0001**	**unknown**	**no**	**negative**	**f**	**52.00**
SRP 1155	***CRB1***	**NM_201253.2**	**c.2812C>T**	**p.(Gln938*)**	**NC_000001.10:g.197398714C>T**	**het**	**AR**	**P**	**PM2, PVS1, PP5**	**-**	**-**	**-**	**unknown**	**no**	**negative**	**f**	**52.00**
SRP 1156	***USH2A***	**NM_206933.2**	**c.13335_13347delinsCTTG**	**p.(Glu4445_Ser4449delinsAspLeu)**	**NC_000001.10:g.215847906_215847918delinsCAAG**	**het**	**AR**	**LP**	**PM2, PM1_SUPPORTING, PP5_STRONG**	**CX104126**	**-**	**-**	**unknown**	**no**	**negative**	**m**	**53.03**
SRP 1156	***USH2A***	**NM_206933.2**	**c.4627+25436_4987+659del**	**p.(?)**	**NC_000001.10:g.216259404_216323160del**	**het**	**AR**	**P**	**PM2, PVS1, PP5**	**na**	**-**	**na**	**unknown**	**no**	**negative**	**m**	**53.03**
SRP 1159	***ABCA4***	**NM_000350.2**	**c.1622T>C**	**p.(Leu541Pro)**	**NC_000001.10:g.94528806A>G**	**het**	**AR**	**P**	**PP3, PM2, PP2, PP5_STRONG, PS4_MODERATE**	**CM990022**	**7901**	**0.0003**	**unknown**	**no**	**negative**	**m**	**52.15**
SRP 1159	***ABCA4***	**NM_000350.2**	**c.3113C>T**	**p.(Ala1038Val)**	**NC_000001.10:g.94508969G>A**	**het**	**AR**	**P**	**PM2, PM1_SUPPORTING, PP2, PP5_STRONG, PS4_MODERATE**	**CM970006**	**7901**	**0.0024**	**unknown**	**no**	**negative**	**m**	**52.15**
SRP 1159	***ABCA4***	**NM_000350.2**	**c.5461-10T>C**	**p.(?)**	**NC_000001.10:g.94476951A>G**	**het**	**AR**	**P**	**PM2, PP5_STRONG, BP4, PS4_MODERATE**	**CS057513**	**1048134**	**0.0002**	**unknown**	**no**	**negative**	**m**	**52.15**
SRP 1159	***ABCA4***	**NM_000350.2**	**c.5603A>T**	**p.(Asn1868Ile)**	**NC_000001.10:g.94476467T>A**	**het**	**AR**	**VUS**	**PP2, PP5, BA1, BS2**	**CM015091**	**1048134**	**0.0426**	**unknown**	**no**	**negative**	**m**	**52.15**
SRP 1161	***PRPF31***	**NM_015629.3**	**c.1275+1G>T**	**p.(?)**	**NC_000019.9:g.54632561G>T**	**het**	**AD**	**LP**	**PM2, PVS1_STRONG**	**-**	**-**	**-**	**unknown**	**no**	**negative**	**f**	**54.55**
SRP 1162	***NR2E3***	**NM_014249.3**	**c.227G>A**	**p.(Arg76Gln)**	**NC_000015.9:g.72103931G>A**	**hom**	**AR+AD**	**LP**	**PM2, PM5, PM1_SUPPORTING, PP5**	**CM000531**	**5530**	**0.0003**	**unknown**	**no**	**negative**	**f**	**46.01**
SRP 1164	***PRPF8***	**NM_006445.3**	**c.6912C>G**	**p.(Phe2304Leu)**	**NC_000017.10:g.1554192G>C**	**het**	**AD**	**P**	**PS1, PP3, PM2, PM1, PP2, PP5**	**CM012448**	**-**	**-**	**de novo**	**no**	**negative**	**m**	**50.37**
SRP 1175	***RP1***	**NM_006269.1**	**c.271del**	**p.(Ser91Alafs*25)**	**NC_000008.10:g.55533797del**	**het**	**AR+AD**	**P**	**PM2, PVS1, PP5**	**-**	**-**	**-**	**unknown**	**no**	**negative**	**m**	**54.60**
SRP 1175	***RP1***	**NM_006269.1**	**c.3157del**	**p.(Tyr1053Thrfs*4)**	**NC_000008.10:g.55539599del**	**het**	**AR+AD**	**P**	**PM2, PVS1, PP5**	**CD004401**	**987374**	**0.000008**	**unknown**	**no**	**negative**	**m**	**54.60**
SRP 1179	***EYS***	**NM_001142800.1**	**c.4045C>T**	**p.(Arg1349*)**	**NC_000006.11:g.65301715G>A**	**het**	**AR**	**P**	**PM2, PVS1, PP5, PS4_MODERATE**	**CM1312776**	**438197**	**0.0001**	**maternal**	**single genes**	**negative**	**m**	**56.42**
SRP 1179	***EYS***	**NM_001142800.1**	**c.4350_4356del**	**p.(Ile1451Profs*3)**	**NC_000006.11:g.65301407_65301413del**	**het**	**AR**	**P**	**PM2, PVS1, PP5**	**CD103602**	**-**	**0.0001**	**paternal**	**single genes**	**negative**	**m**	**56.42**
SRP 1181	***NR2E3***	**NM_014249.3**	**c.263G>T**	**p.(Gly88Val)**	**NC_000015.9:g.72104123G>T**	**het**	**AR+AD**	**LP**	**PM2, PM5, PM1_SUPPORTING, PP5**	**CM043307**	**987298**	**0.0000**	**unknown**	**no**	**negative**	**m**	**53.54**
SRP 1181	***NR2E3***	**NM_014249.3**	**c.406G>T**	**p.(Glu136*)**	**NC_000015.9:g.72104351G>T**	**het**	**AR+AD**	**P**	**PM2, PVS1, PP5**	**CM2036724**	**813198**	**-**	**unknown**	**no**	**negative**	**m**	**53.54**
SRP 1184	***RPGR***	**NM_001034853.1**	**c.1836del**	**p.(Asn612Lysfs*2)**	**NC_000023.10:g.38146416del**	**hemi**	**XLR+XLD**	**P**	**PM2, PVS1, PP5**	**-**	**-**	**-**	**de novo**	**no**	**negative**	**m**	**51.99**
SRP 1185	***PDE6A***	**NM_000440.2**	**c.304C>A**	**p.(Arg102Ser)**	**NC_000005.9:g.149323933G>T**	**hom**	**AR**	**P**	**PM2, PM5, PP5_STRONG**	**CM994742**	**193099**	**0.0002**	**unknown**	**no**	**unknown**	**m**	**52.46**
SRP 1186	***ABCA4***	**NM_000350.2**	**c.2588G>C**	**p.(Gly863Ala)**	**NC_000001.10:g.94517254C>G**	**het**	**AR**	**P**	**PP3, PM2, PP2, PP5_STRONG, PS4_MODERATE**	**CM970003&CS024003**	**-**	**0.0044**	**unknown**	**no**	**unknown**	**m**	**43.81**
SRP 1186	***ABCA4***	**NM_000350.2**	**c.5603A>T**	**p.(Asn1868Ile)**	**NC_000001.10:g.94476467T>A**	**het**	**AR**	**VUS**	**PP2, PP5, BA1, BS2**	**CM015091**	**-**	**0.0426**	**unknown**	**no**	**unknown**	**m**	**43.81**
SRP 1186	*USH2A*	NM_206933.2	c.2299del	p.(Glu767Serfs*21)	NC_000001.10:g.216420437del	het	AR	P	PM2, PVS1, PP5, PS4	CD982997	-	0.0008	unknown	no	unknown	m	43.81
SRP 1186	*USH2A*	NM_206933.2	c.4714C>T	p.(Leu1572Phe)	NC_000001.10:g.216270469G>A	het	AR	VUS	PP3, PM2, BP6_STRONG	CM119884	-	0.0008	unknown	no	unknown	m	43.81
SRP 1186	*USH2A*	NM_206933.2	c.6163G>A	p.(Ala2055Thr)	NC_000001.10:g.216221876C>T	het	AR	VUS	PM2	-	-	0.00002	unknown	no	unknown	m	43.81
SRP 1196	***MERTK***	**NM_006343.2**	**c.1689_1690+5delinsATATTA**	**p.(?)**	**NC_000002.11:g.112758862_112758868delinsATATTA**	**hom**	**AR**	**P**	**PM2, PVS1, PP5**	**CX2036907**	**-**	**-**	**unknown**	**no**	**negative**	**f**	**42.94**
SRP 1197	***HK1***	**NM_000188.2**	**c.2539G>A**	**p.(Glu847Lys)**	**NC_000010.10:g.71158514G>A**	**het**	**AR+AD**	**LP**	**PP3, PM2, PP2, PP5_STRONG**	**CM1412146**	**-**	**0.0001**	**unknown**	**no**	**negative**	**m**	**57.06**
SRP 1197	*NR2E3*	NM_014249.3	c.481del	p.(Thr161Hisfs*18)	NC_000015.9:g.72104426del	het	AR+AD	P	PM2, PVS1, PP5	CD043411	-	0.0002	unknown	no	negative	m	57.06
SRP 1198	***CFAP410***	**NM_004928.2**	**c.443_444del**	**p.(Glu148Glyfs*21)**	**NC_000021.8:g.45751835_45751836del**	**het**	**AR**	**P**	**PM2, PVS1, PP5**	**-**	**-**	**0.0001**	**unknown**	**no**	**negative**	**m**	**53.97**
SRP 1198	***CFAP410***	**NM_004928.2**	**c.59_62del**	**p.(Val20Alafs*24)**	**NC_000021.8:g.45759019_45759022del**	**het**	**AR**	**P**	**PM2, PVS1, PP5**	**-**	**-**	**-**	**unknown**	**no**	**negative**	**m**	**53.97**
SRP 1199	***TULP1***	**NM_003322.3**	**c.1445G>A**	**p.(Arg482Gln)**	**NC_000006.11:g.35467808C>T**	**het**	**AR**	**LP**	**PP3, PM2, PM5, PP5**	**CM123565**	**-**	**0.0001**	**maternal**	**no**	**negative**	**m**	**47.26**
SRP 1199	***TULP1***	**NM_003322.3**	**c.1163C>A**	**p.(Pro388Gln)**	**NC_000006.11:g.35471575G>T**	**het**	**AR**	**LP**	**PP3, PM2, PM1_SUPPORTING, PM3**	**-**	**-**	**0.000007**	**paternal**	**no**	**negative**	**m**	**47.26**
SRP 1199	*ARL2BP*	NM_012106.3	c.294-1G>C	p.(?)	NC_000016.9:g.57284322G>C	het	AR	LP	PM2, PVS1	-	-	0.000004	unknown	no	negative	m	47.26
SRP 1200	***USH2A***	**NM_206933.2**	**c.12232G>T**	**p.(Glu4078*)**	**NC_000001.10:g.215853553C>A**	**het**	**AR**	**P**	**PM2, PVS1, PP5**	**CM165550**	**-**	**0.0001**	**unknown**	**no**	**negative**	**f**	**46.63**
SRP 1200	***USH2A***	**NM_206933.2**	**c.2276G>T**	**p.(Cys759Phe)**	**NC_000001.10:g.216420460C>A**	**het**	**AR**	**P**	**PP3, PM2, PM1_SUPPORTING, PP5_STRONG, PS4_MODERATE**	**CM001372**	**2356**	**0.0010**	**unknown**	**no**	**negative**	**f**	**46.63**
SRP 1201	***USH2A***	**NM_206933.2**	**c.100C>T**	**p.(Arg34*)**	**NC_000001.10:g.216595579G>A**	**het**	**AR**	**P**	**PM2, PVS1, PP5, PS4**	**CM001803**	**-**	**0.0001**	**unknown**	**no**	**negative**	**f**	**46.80**
SRP 1201	***USH2A***	**NM_206933.2**	**c.2276G>T**	**p.(Cys759Phe)**	**NC_000001.10:g.216420460C>A**	**het**	**AR**	**P**	**PP3, PM2, PM1_SUPPORTING, PP5_STRONG, PS4_MODERATE**	**CM001372**	**2356**	**0.0010**	**unknown**	**no**	**negative**	**f**	**46.80**
SRP 1203	***RP1***	**NM_006269.1**	**c.1975A>T**	**p.(Lys659*)**	**NC_000008.10:g.55538417A>T**	**het**	**AR+AD**	**P**	**PM2, PVS1_STRONG, PP5_STRONG**	**-**	**-**	**-**	**unknown**	**no**	**negative**	**f**	**58.00**
SRP 1204	***CWC27***	**NM_005869.2**	**c.666_669del**	**p.(Ser222Argfs*3)**	**NC_000005.9:g.64084844_64084847del**	**het**	**AR**	**P**	**PM2, PVS1, PP5**	**-**	**-**	**-**	**unknown**	**no**	**negative**	**m**	**53.29**
SRP 1204	***CWC27***	**NM_005869.2**	**na**	**na**	**NC_000005.9:g.63985369_64118651delins[CTGG;63985092_63985116;GGTCATG;63985099_63985162]**	**het**	**AR**	**LP**	**PM2, PVS1**	**na**	**-**	**na**	**maternal**	**no**	**negative**	**m**	**53.29**
SRP 1204	*RP11*	NM_178857.5	c.326_327insT	p.(Lys111Glnfs*27)	NC_000008.10:g.10480385_10480386insA	het	AD+AR	P	PM2, PVS1, PP5	CI1474566	-	0.0019	maternal	no	negative	m	53.29
SRP 1204	*RP11*	NM_178857.5	c.5884G>C	p.(Glu1962Gln)	NC_000008.10:g.10465724C>G	het	AD+AR	VUS	PM2, BP4	-	-	-	unknown	no	negative	m	53.29
SRP 1204	*SAG*	NM_000541.4	c.577C>T	p.(Arg193*)	NC_000002.11:g.234237188C>T	het	AR	P	PM2, PVS1, PP5	CM981785	-	0.0003	unknown	no	negative	m	53.29
SRP 1209	***BEST1***	**NM_004183.3**	**c.287_298del**	**p.(Gln96_Asn99del)**	**NC_000011.9:g.61723229_61723240del**	**hom**	**AR+AD**	**P**	**PM2, PM4, PM1, PP5_STRONG, PS4_MODERATE**	**CD2017335**	**-**	**-**	**unknown**	**no**	**negative**	**m**	**54.71**
SRP 1211	***ABCA4***	**NM_000350.2**	**c.5603A>T**	**p.(Asn1868Ile)**	**NC_000001.10:g.94476467T>A**	**het**	**AR**	**VUS**	**PP2, PP5, BA1, BS2**	**CM015091**	**-**	**0.0426**	**unknown**	**no**	**negative**	**f**	**45.64**
SRP 1211	***ABCA4***	**NM_000350.2**	**c.5714+5G>A**	**p.(?)**	**NC_000001.10:g.94476351C>T**	**het**	**AR**	**P**	**PP3, PM2, PP5_STRONG, PS4_MODERATE**	**CS982057**	**-**	**0.0003**	**unknown**	**no**	**negative**	**f**	**45.64**
SRP 1211	*ABCA4*	NM_000350.2	c.2105T>C	p.(Leu702Pro)	NC_000001.10:g.94526148A>G	het	AR	VUS	PP3, PM2, PP2	-	-	-	unknown	no	negative	f	45.64
SRP 1215	***EYS***	**NM_001142800.1**	**c.4350_4356del**	**p.(Ile1451Profs*3)**	**NC_000006.11:g.65301407_65301413del**	**het**	**AR**	**P**	**PM2, PVS1, PP5**	**CD103602**	**195936**	**0.0001**	**unknown**	**no**	**negative**	**f**	**50.33**
SRP 1215	***EYS***	**NM_001142800.1**	**c.6416G>A**	**p.(Cys2139Tyr)**	**NC_000006.11:g.64940493C>T**	**het**	**AR**	**P**	**PP3, PM2, PM5, PP5_STRONG**	**CM102730**	**189230**	**0.0001**	**unknown**	**no**	**negative**	**f**	**50.33**
SRP 1216	***RP1***	**NM_006269.1**	**c.2374A>T**	**p.(Lys792*)**	**NC_000008.10:g.55538816A>T**	**het**	**AR+AD**	**P**	**PM2, PVS1_STRONG, PP5_STRONG**	**CM1919457**	**853663**	**0.0001**	**unknown**	**no**	**negative**	**m**	**55.24**
SRP 1217	***PRPF31***	**NM_015629.3**	**c.1146+2T>G**	**p.(?)**	**NC_000019.9:g.54631754T>G**	**het**	**AD**	**P**	**PM2, PVS1, PP5**	**-**	**-**	**-**	**unknown**	**no**	**negative**	**f**	**34.52**
SRP 1220	***CHM***	**NM_000390.2**	**c.715C>T**	**p.(Arg239*)**	**NC_000023.10:g.85213970G>A**	**het**	**XLD**	**P**	**PM2, PVS1, PP5**	**CM010795**	**-**	**0.0001**	**unknown**	**no**	**negative**	**f**	**44.96**
SRP 1225	***RHO***	**NM_000539.3**	**c.937-1G>C**	**p.(?)**	**NC_000003.11:g.129252450G>C**	**het**	**AR+AD**	**P**	**PM2, PVS1_STRONG, PP5_STRONG**	**-**	**-**	**-**	**unknown**	**no**	**negative**	**f**	**46.00**
SRP 1227	***MERTK***	**NM_006343.2**	**c.2163T>A**	**p.(His721Gln)**	**NC_000002.11:g.112777073T>A**	**hom**	**AR**	**P**	**PP3, PM2, PM5, PM1, PP5, PP5_STRONG**	**CM1811516**	**866882**	**0.0001**	**unknown**	**no**	**negative**	**m**	**51.77**
SRP 1231	***FAM161A***	**NM_001201543.1**	**c.1460del**	**p.(Arg487Leufs*13)**	**NC_000002.11:g.62066679del**	**hom**	**AR**	**P**	**PM2, PVS1, PP5**	**-**	**-**	**-**	**paternal/maternal**	**no**	**negative**	**m**	**51.59**
SRP 1232	***RPE65***	**NM_000329.2**	**c.11+5G>A**	**p.(?)**	**NC_000001.10:g.68915573C>T**	**het**	**AR**	**P**	**PP3, PM2, PP5_STRONG, PS4_MODERATE**	**CS971898**	**-**	**0.0001**	**unknown**	**no**	**negative**	**f**	**58.79**
SRP 1232	***RPE65***	**NM_000329.2**	**c.1102T>C**	**p.(Tyr368His)**	**NC_000001.10:g.68903896A>G**	**het**	**AR**	**LP**	**PP3, PM2, PP2, PP5_STRONG**	**CM021674**	**29870**	**0.0001**	**unknown**	**no**	**negative**	**f**	**58.79**
SRP 1236	***SPATA7***	**NM_018418.4**	**c.1183C>T**	**p.(Arg395*)**	**NC_000014.8:g.88903909C>T**	**het**	**AR**	**P**	**PM2, PVS1_STRONG, PP5_STRONG**	**CM091507**	**-**	**0.0001**	**unknown**	**no**	**negative**	**f**	**45.72**
SRP 1236	***SPATA7***	**NM_018418.4**	**c.3G>A**	**p.(Met1?)**	**NC_000014.8:g.88852165G>A**	**het**	**AR**	**LP**	**PM2, PVS1_MODERATE, PP5_STRONG**	**CM124907**	**844972**	**0.0001**	**unknown**	**no**	**negative**	**f**	**45.72**
SRP 1240	***USH2A***	**NM_206933.2**	**c.2276G>T**	**p.(Cys759Phe)**	**NC_000001.10:g.216420460C>A**	**het**	**AR**	**P**	**PP3, PM2, PM1_SUPPORTING, PP5_STRONG, PS4_MODERATE**	**CM001372**	**2356**	**0.0010**	**unknown**	**no**	**negative**	**m**	**60.98**
SRP 1240	***USH2A***	**NM_206933.2**	**c.8917_8918del**	**p.(Leu2973Lysfs*79)**	**NC_000001.10:g.216019307_216019308del**	**het**	**AR**	**P**	**PM2, PVS1, PP5**	**CD153043**	**-**	**-**	**unknown**	**no**	**negative**	**m**	**60.98**
SRP 1242	***IQCB1***	**NM_001023570.2**	**c.1518_1519del**	**p.(His506Glnfs*13)**	**NC_000003.11:g.121491454_121491455del**	**het**	**AR**	**P**	**PM2, PVS1_STRONG, PP5_STRONG**	**CD050887**	**-**	**0.0001**	**unknown**	**no**	**negative**	**m**	**51.78**
SRP 1242	***IQCB1***	**NM_001023570.2**	**c.424_425del**	**p.(Phe142Profs*5)**	**NC_000003.11:g.121527828_121527829del**	**het**	**AR**	**P**	**PM2, PVS1, PP5, PS4**	**CD050884**	**1831**	**0.0001**	**unknown**	**no**	**negative**	**m**	**51.78**
SRP 1246	***PDE6B***	**NM_000283.3**	**c.739T>A**	**p.(Phe247Ile)**	**NC_000004.11:g.647668T>A**	**hom**	**AR**	**LP**	**PM2, PP5_STRONG, PS4_MODERATE**	**CM175551**	**349362**	**0.0001**	**unknown**	**single genes**	**negative**	**m**	**53.27**
SRP 1246	*GNAT1*	NM_144499.2	c.514C>G	p.(Arg172Gly)	NC_000003.11:g.50231250C>G	het	AR+AD	VUS	PM2	-	-	-	unknown	single genes	negative	m	53.27
SRP 1247	***PRPF8***	**NM_006445.3**	**c.6991G>A**	**p.(Glu2331Lys)**	**NC_000017.10:g.1554113C>T**	**het**	**AD**	**P**	**PP3, PM2, PM1, PP2, PP5_STRONG**	**-**	**-**	**-**	**unknown**	**single genes**	**negative**	**m**	**56.99**
SRP 1253	***PRPF31***	**NM_015629.3**	**c.238+705_1375-727del**	**p.(?)**	**NC_000019.9:g.54622718_54634011del**	**het**	**AD**	**LP**	**PM2, PVS1**	**na**	**-**	**na**	**unknown**	**single genes**	**negative**	**f**	**53.29**
SRP 1253	*RIMS1*	NM_014989.4	c.1626G>C	p.(Glu542Asp)	NC_000006.11:g.72892800G>C	het	AD	VUS	PM2, BP4	-	-	0.0001	unknown	single genes	negative	f	53.29
SRP 1254	***PRPF31***	**NM_015629.3**	**c.896G>A**	**p.(Cys299Tyr)**	**NC_000019.9:g.54629943G>A**	**het**	**AD**	**LP**	**PP3, PM2, PM5, PP2**	**CM1812263**	**-**	**-**	**unknown**	**single genes**	**negative**	**f**	**58.73**
SRP 1264	***HK1***	**NM_000188.2**	**c.1334C>T**	**p.(Ser445Leu)**	**NC_000010.10:g.71142311C>T**	**het**	**AD**	**P**	**PP3, PM2, PP2, PP5_STRONG**	**CM1910439**	**421007**	**-**	**unknown**	**no**	**unknown**	**m**	**35.04**
SRP 1264	*GUCY2D*	NM_000180.3	c.826del	p.(Leu276Cysfs*119)	NC_000017.10:g.7907274del	het	AR+AD	P	PM2, PVS1, PP5	-	1184899	-	unknown	no	unknown	m	35.04
SRP 1267	***PRPF8***	**NM_006445.3**	**c.5804G>A**	**p.(Arg1935His)**	**NC_000017.10:g.1558827C>T**	**het**	**AD**	**P**	**PS1_MODERATE, PP3, PM2, PM5_SUPPORTING, PP2, PP5_STRONG**	**CM149178**	**546782**	**-**	**unknown**	**no**	**negative**	**f**	**37.20**
SRP 1270	***USH2A***	**NM_206933.2**	**c.11105G>A**	**p.(Trp3702*)**	**NC_000001.10:g.215933128C>T**	**het**	**AR**	**P**	**PM2, PVS1, PP5**	**CM140502**	**374674**	**0.000007**	**unknown**	**no**	**negative**	**f**	**39.62**
SRP 1270	***USH2A***	**NM_206933.2**	**c.12575G>A**	**p.(Arg4192His)**	**NC_000001.10:g.215848678C>T**	**het**	**AR**	**P**	**PM2, PP5_STRONG, PS4_MODERATE**	**CM108280**	**-**	**0.0006**	**unknown**	**no**	**negative**	**f**	**39.62**
SRP 1270	*RP11*	NM_178857.5	c.5959C>T	p.(Gln1987*)	NC_000008.10:g.10465649G>A	het	AR+AD	P	PM2, PVS1_STRONG, PP5_STRONG	CM120689	-	0.0016	unknown	no	negative	f	39.62
SRP 1277	***RPGRIP1***	**NM_020366.3**	**c.2367+23del**	**p.(?)**	**NC_000014.8:g.21793565del**	**het**	**AR**	**LP**	**PM2, PP5_STRONG, BP7**	**CD174461**	**-**	**0.0002**	**unknown**	**no**	**negative**	**m**	**39.60**
SRP 1277	***RPGRIP1***	**NM_020366.3**	**c.3340-1072_3533-254del**	**p.(?)**	**NC_000014.8:g.21810123_21813018del**	**het**	**AR**	**LP**	**PM2, PVS1**	**na**	**-**	**na**	**unknown**	**no**	**negative**	**m**	**39.60**
SRP 1277	*RPGRIP1*	NM_020366.3	c.268G>A	p.(Val90Ile)	NC_000014.8:g.21769174G>A	het	AR	VUS	PM2, PP5, BP4	CM2036786	-	0.0001	unknown	no	negative	m	39.60
SRP 1277	*OTX2*	NM_021728.3	c.125C>T	p.(Thr42Ile)	NC_000014.8:g.57271054G>A	het	AD	VUS	PP3, PM2, PP2	-	-	-	unknown	no	negative	m	39.60
SRP 1278	***PRPH2***	**NM_000322.4**	**c.911_912dup**	**p.(Gly305Argfs*20)**	**NC_000006.11:g.42666162_42666163dup**	**het**	**AD**	**P**	**PM2, PVS1_STRONG, PP5_STRONG**	**-**	**1275765**	**-**	**unknown**	**no**	**negative**	**f**	**42.29**
SRP 1279	***CRB1***	**NM_201253.2**	**c.134_135del**	**p.(Cys45*)**	**NC_000001.10:g.197297615_197297616del**	**het**	**AR**	**P**	**PM2, PVS1, PP5**	**-**	**1275767**	**0.000004**	**unknown**	**no**	**negative**	**f**	**38.85**
SRP 1279	***CRB1***	**NM_201253.2**	**c.2290C>T**	**p.(Arg764C

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STGD 396	***ABCA4***	**NM_000350.2**	**c.1648G>A**	**p.(Gly550Arg)**	**NC_000001.10:g.94528780C>T**	**hom**	**AR**	**P**	**PP3, PM2, PP2, PP5_STRONG**	**CM013504**	**-**	**0.0001**	**unknown**	**no**	**negative**	**f**	**49.55**
STGD 397	***ABCA4***	**NM_000350.2**	**c.5882G>A**	**p.(Gly1961Glu)**	**NC_000001.10:g.94473807C>T**	**het**	**AR**	**P**	**PP3, PM2, PM5, PP2, PP5_STRONG, PS4_MODERATE**	**CM970016**	**635154**	**0.0047**	**maternal**	**single genes**	**negative**	**m**	**48.84**
STGD 397	***ABCA4***	**NM_000350.2**	**na**	**na**	**NC_000001.10:g.94452244_94471761del**	**het**	**AR**	**LP**	**PM2, PVS1**	**na**	**-**	**na**	**paternal**	**single genes**	**negative**	**m**	**48.84**
STGD 398	***ABCA4***	**NM_000350.2**	**c.5642C>T**	**p.(Ala1881Val)**	**NC_000001.10:g.94476428G>A**	**het**	**AR**	**P**	**PM2, PM5_SUPPORTING, PM1, PP2, PP5_STRONG**	**CM1210005**	**853893**	**0.0001**	**unknown**	**no**	**negative**	**m**	**45.58**
STGD 398	***ABCA4***	**NM_000350.2**	**c.6215G>A**	**p.(Ser2072Asn)**	**NC_000001.10:g.94467481C>T**	**het**	**AR**	**P**	**PP3, PM2, PM1, PP2, PP5_STRONG**	**CM150888**	**-**	**-**	**unknown**	**no**	**negative**	**m**	**45.58**
STGD 398	***ABCA4***	**NM_000350.2**	**c.868C>T**	**p.(Arg290Trp)**	**NC_000001.10:g.94546265G>A**	**het**	**AR**	**P**	**PM2, PM5, PM1_SUPPORTING, PP2, PP5_STRONG**	**CM014287**	**-**	**0.0001**	**unknown**	**no**	**negative**	**m**	**45.58**
STGD 399	***ABCA4***	**NM_000350.2**	**c.1834C>T**	**p.(Gln612*)**	**NC_000001.10:g.94528236G>A**	**het**	**AR**	**P**	**PM2, PVS1, PP5**	**CM1517305**	**-**	**-**	**unknown**	**no**	**negative**	**m**	**53.42**
STGD 399	***ABCA4***	**NM_000350.2**	**c.5882G>A**	**p.(Gly1961Glu)**	**NC_000001.10:g.94473807C>T**	**het**	**AR**	**P**	**PP3, PM2, PM5, PP2, PP5_STRONG, PS4_MODERATE**	**CM970016**	**635154**	**0.0047**	**unknown**	**no**	**negative**	**m**	**53.42**
STGD 402	***ABCA4***	**NM_000350.2**	**c.4786dup**	**p.(Arg1596Lysfs*5)**	**NC_000001.10:g.94487258dup**	**het**	**AR**	**P**	**PM2, PVS1, PP5**	**CI2036592**	**813138**	**-**	**shared with sister**	**no**	**negative**	**f**	**46.80**
STGD 402	***ABCA4***	**NM_000350.2**	**c.5603A>T**	**p.(Asn1868Ile)**	**NC_000001.10:g.94476467T>A**	**het**	**AR**	**VUS**	**PP2, PP5, BA1, BS2**	**CM015091**	**-**	**0.0426**	**maternal**	**no**	**negative**	**f**	**46.80**
STGD 405	***ABCA4***	**NM_000350.2**	**c.5603A>T**	**p.(Asn1868Ile)**	**NC_000001.10:g.94476467T>A**	**het**	**AR**	**VUS**	**PP2, PP5, BA1, BS2**	**CM015091**	**-**	**0.0426**	**unknown**	**no**	**negative**	**f**	**54.23**
STGD 405	***ABCA4***	**NM_000350.2**	**c.5942C>G**	**p.(Thr1981Arg)**	**NC_000001.10:g.94473253G>C**	**het**	**AR**	**P**	**PP3, PM2, PM1, PP2, PP5_STRONG**	**CM171201**	**-**	**-**	**unknown**	**no**	**negative**	**f**	**54.23**
STGD 408	***ABCA4***	**NM_000350.2**	**c.571-1G>T**	**p.(?)**	**NC_000001.10:g.94564548C>A**	**het**	**AR**	**P**	PM2, PVS1_STRONG, PP5_STRONG	**CS013074**	**-**	**-**	**maternal**	**no**	**negative**	**m**	**52.52**
STGD 408	***ABCA4***	**NM_000350.2**	**c.634C>T**	**p.(Arg212Cys)**	**NC_000001.10:g.94564484G>A**	**het**	**AR**	**P**	**PP3, PM2, PP2, PP5_STRONG, PS4_MODERATE**	**CM980004**	**7898**	**0.0001**	**paternal**	**no**	**negative**	**m**	**52.52**
STGD 408	*OPN1LW*	NM_020061.4	c.532A>G	p.(Ile178Val)	NC_000023.10:g.153418535A>G	hemi	XLR	B	BS1, BS2, BP4, BP6_STRONG	CM890024	-	0.0214	maternal	no	negative	m	52.52
STGD 408	*OPN1LW*	NM_020061.4	c.538T>G	p.(Ser180Ala)	NC_000023.10:g.153418541T>G	hemi	XLR	B	BA1, BS2, BP6	CM045803	-	0.2244	maternal	no	negative	m	52.52
STGD 408	*OPN1MW*	NM_000513.1	c.532A>G	p.(Ile178Val)	NC_000023.10:g.153455665A>G	het	XLR	B	BA1, BS1, BS2, BP4	CM890024	-	0.0214	maternal	no	negative	m	52.52
STGD 409	***ABCA4***	**NM_000350.2**	**c.1622T>C**	**p.(Leu541Pro)**	**NC_000001.10:g.94528806A>G**	**het**	**AR**	**P**	**PP3, PM2, PP2, PP5_STRONG, PS4_MODERATE**	**CM990022**	**7901**	**0.0003**	**unknown**	**no**	**negative**	**f**	**50.34**
STGD 409	***ABCA4***	**NM_000350.2**	**c.3113C>T**	**p.(Ala1038Val)**	**NC_000001.10:g.94508969G>A**	**het**	**AR**	**P**	**PM2, PM1_SUPPORTING, PP2, PP5_STRONG, PS4_MODERATE**	**CM970006**	**7901**	**0.0024**	**unknown**	**no**	**negative**	**f**	**50.34**
STGD 409	***ABCA4***	**NM_000350.2**	**c.5882G>A**	**p.(Gly1961Glu)**	**NC_000001.10:g.94473807C>T**	**het**	**AR**	**P**	**PP3, PM2, PM5, PP2, PP5_STRONG, PS4_MODERATE**	**CM970016**	**635154**	**0.0047**	**unknown**	**no**	**negative**	**f**	**50.34**
STGD 411	***ABCA4***	**NM_000350.2**	**c.1253T>C**	**p.(Phe418Ser)**	**NC_000001.10:g.94544249A>G**	**het**	**AR**	**P**	**PP3, PM2, PP2, PP5_STRONG**	**CM117250**	**193580**	**0.0001**	**paternal**	**no**	**negative**	**f**	**54.11**
STGD 411	***ABCA4***	**NM_000350.2**	**c.2626C>T**	**p.(Gln876*)**	**NC_000001.10:g.94517216G>A**	**het**	**AR**	**P**	**PM2, PVS1, PP5**	**CM042263**	**-**	**-**	**unknown**	**no**	**negative**	**f**	**54.11**
STGD 411	*RP11*	NM_178857.5	c.7037A>C	p.(Glu2346Ala)	NC_000008.10:g.10464571T>G	het	AD+AR	VUS	PM2, BP4	-	-	-	unknown	no	negative	f	54.11
STGD 414	***ABCA4***	**NM_000350.2**	**c.2829del**	**p.(Pro944Glnfs*6)**	**NC_000001.10:g.94512565del**	**hom**	**AR**	**P**	**PM2, PVS1, PP5**	**CD1310357**	**987414**	**-**	**unknown**	**no**	**negative**	**f**	**47.43**
STGD 416	***ABCA4***	**NM_000350.2**	**c.3413T>A**	**p.(Leu1138His)**	**NC_000001.10:g.94506874A>T**	**het**	**AR**	**P**	**PP3, PM2, PM5_SUPPORTING, PM1, PP2, PP5_STRONG**	**CM1824747**	**-**	**-**	**unknown**	**no**	**negative**	**m**	**45.65**
STGD 416	***ABCA4***	**NM_000350.2**	**c.5714+5G>A**	**p.(?)**	**NC_000001.10:g.94476351C>T**	**het**	**AR**	**P**	**PP3, PM2, PP5_STRONG, PS4_MODERATE**	**CS982057**	**1048131**	**0.0003**	**unknown**	**no**	**negative**	**m**	**45.65**
STGD 419	***ABCA4***	**NM_000350.2**	**c.3856_3859del**	**p.(Phe1286Argfs*102)**	**NC_000001.10:g.94502301_94502304del**	**het**	**AR**	**P**	**PM2, PVS1, PP5**	**-**	**-**	**-**	**unknown**	**no**	**negative**	**m**	**56.49**
STGD 419	***ABCA4***	**NM_000350.2**	**c.5882G>A**	**p.(Gly1961Glu)**	**NC_000001.10:g.94473807C>T**	**het**	**AR**	**P**	**PP3, PM2, PM5, PP2, PP5_STRONG, PS4_MODERATE**	**CM970016**	**635154**	**0.0047**	**unknown**	**no**	**negative**	**m**	**56.49**
STGD 423	***ABCA4***	**NM_000350.2**	**c.4253+43G>A**	**p.(?)**	**NC_000001.10:g.94496509C>T**	**het**	**AR**	**LP**	**PM2, PP5_STRONG, BP7**	**CS193151**	**-**	**0.0047**	**unknown**	**no**	**negative**	**f**	**39.94**
STGD 423	***ABCA4***	**NM_000350.2**	**c.6098T>C**	**p.(Leu2033Pro)**	**NC_000001.10:g.94471046A>G**	**het**	**AR**	**LP**	**PP3, PM2, PM5_SUPPORTING, PM1, PP2, PP5, PS4_MODERATE**	**CM175571**	**-**	**-**	**unknown**	**no**	**negative**	**f**	**39.94**
STGD 427	***ABCA4***	**NM_000350.2**	**c.5196+1G>A**	**p.(?)**	**NC_000001.10:g.94485137C>T**	**het**	**AR**	**P**	**PM2, PVS1, PP5**	**CS971603**	**-**	**0.0001**	**unknown**	**no**	**negative**	**f**	**48.15**
STGD 427	***ABCA4***	**NM_000350.2**	**c.5882G>A**	**p.(Gly1961Glu)**	**NC_000001.10:g.94473807C>T**	**het**	**AR**	**P**	**PP3, PM2, PM5, PP2, PP5_STRONG, PS4_MODERATE**	**CM970016**	**635154**	**0.0047**	**unknown**	**no**	**negative**	**f**	**48.15**
STGD 427	*FBN2*	NM_001999.3	c.8058C>A	p.(Phe2686Leu)	NC_000005.9:g.127599251G>T	het	AD	VUS	PM2	-	-	0.0001	unknown	no	negative	f	48.15
STGD 427	*SEMA4A*	NM_022367.3	c.305C>T	p.(Pro102Leu)	NC_000001.10:g.156127865C>T	het	AD	VUS	PM2, BP4	-	-	0.0001	unknown	no	negative	f	48.15
STGD 428	***ABCA4***	**NM_000350.2**	**c.52C>T**	**p.(Arg18Trp)**	**NC_000001.10:g.94586550G>A**	**het**	**AR**	**P**	**PP3, PM2, PM5, PP2, PP5_STRONG**	**CM980003**	**1048166**	**0.0001**	**unknown**	**no**	**negative**	**f**	**46.46**
STGD 428	***ABCA4***	**NM_000350.2**	**c.5882G>A**	**p.(Gly1961Glu)**	**NC_000001.10:g.94473807C>T**	**het**	**AR**	**P**	**PP3, PM2, PM5, PP2, PP5_STRONG, PS4_MODERATE**	**CM970016**	**635154**	**0.0047**	**unknown**	**no**	**negative**	**f**	**46.46**
STGD 429	***ABCA4***	**NM_000350.2**	**c.2588G>C**	**p.(Gly863Ala)**	**NC_000001.10:g.94517254C>G**	**het**	**AR**	**P**	**PP3, PM2, PP2, PP5_STRONG, PS4_MODERATE**	**CM970003&CS024003**	**1048132**	**0.0044**	**unknown**	**no**	**negative**	**m**	**46.59**
STGD 429	***ABCA4***	**NM_000350.2**	**c.5603A>T**	**p.(Asn1868Ile)**	**NC_000001.10:g.94476467T>A**	**het**	**AR**	**VUS**	**PP2, PP5, BA1, BS2**	**CM015091**	**1048132**	**0.0426**	**unknown**	**no**	**negative**	**m**	**46.59**
STGD 429	***ABCA4***	**NM_000350.2**	**c.5929G>A**	**p.(Gly1977Ser)**	**NC_000001.10:g.94473266C>T**	**het**	**AR**	**P**	**PP3, PM2, PM5, PM1, PP2, PP5_STRONG**	**CM990069**	**-**	**0.0001**	**unknown**	**no**	**negative**	**m**	**46.59**
STGD 430	***ABCA4***	**NM_000350.2**	**c.5882G>A**	**p.(Gly1961Glu)**	**NC_000001.10:g.94473807C>T**	**het**	**AR**	**P**	**PP3, PM2, PM5, PP2, PP5_STRONG, PS4_MODERATE**	**CM970016**	**635154**	**0.0047**	**unknown**	**no**	**negative**	**f**	**49.24**
STGD 430	***ABCA4***	**NM_000350.2**	**c.1817G>A**	**p.(Gly606Asp)**	**NC_000001.10:g.94528253C>T**	**het**	**AR**	**LP**	**PP3, PM2, PM1, PP2**	**CM070631**	**-**	**0.0001**	**unknown**	**no**	**negative**	**f**	**49.24**
STGD 430	*RAX2*	NM_032753.3	c.76A>C	p.(Lys26Gln)	NC_000019.9:g.3771665T>G	het	AD	VUS	PM2, BP6	-	-	0.0001	unknown	no	negative	f	49.24
STGD 431	***ABCA4***	**NM_000350.2**	**c.122G>A**	**p.(Trp41*)**	**NC_000001.10:g.94578567C>T**	**het**	**AR**	**P**	**PM2, PVS1, PP5**	**CM032795**	**-**	**0.0001**	**unknown**	**no**	**negative**	**m**	**51.02**
STGD 431	***ABCA4***	**NM_000350.2**	**c.2588G>C**	**p.(Gly863Ala)**	**NC_000001.10:g.94517254C>G**	**het**	**AR**	**P**	**PP3, PM2, PP2, PP5_STRONG, PS4_MODERATE**	**CM970003&CS024003**	**-**	**0.0044**	**unknown**	**no**	**negative**	**m**	**51.02**
STGD 431	***ABCA4***	**NM_000350.2**	**c.5603A>T**	**p.(Asn1868Ile)**	**NC_000001.10:g.94476467T>A**	**het**	**AR**	**VUS**	**PP2, PP5, BA1, BS2**	**CM015091**	**-**	**0.0426**	**unknown**	**no**	**negative**	**m**	**51.02**
STGD 435	***ABCA4***	**NM_000350.2**	**c.5318C>T**	**p.(Ala1773Val)**	**NC_000001.10:g.94480241G>A**	**hom**	**AR**	**P**	**PP3, PM2, PM5, PM1, PP2, PP5_STRONG**	**CM087708**	**-**	**0.0001**	**unknown**	**no**	**negative**	**f**	**51.21**
STGD 436	***ABCA4***	**NM_000350.2**	**c.3051-1G>C**	**p.(?)**	**NC_000001.10:g.94509032C>G**	**het**	**AR**	**P**	**PM2, PVS1, PP5**	**-**	**-**	**-**	**unknown**	**no**	**negative**	**f**	**58.63**
STGD 436	***ABCA4***	**NM_000350.2**	**c.5882G>A**	**p.(Gly1961Glu)**	**NC_000001.10:g.94473807C>T**	**het**	**AR**	**P**	**PP3, PM2, PM5, PP2, PP5_STRONG, PS4_MODERATE**	**CM970016**	**635154**	**0.0047**	**unknown**	**no**	**negative**	**f**	**58.63**
STGD 436	6p21.1	na	na	na	NC_000006.11:g.42039467_42275673dup	het	na	LP	PM2, PVS1	na	-	na	unknown	no	negative	f	58.63
STGD 437	***ABCA4***	**NM_000350.2**	**c.5882G>A**	**p.(Gly1961Glu)**	**NC_000001.10:g.94473807C>T**	**het**	**AR**	**P**	**PP3, PM2, PM5, PP2, PP5_STRONG, PS4_MODERATE**	**CM970016**	**635154**	**0.0047**	**unknown**	**no**	**negative**	**f**	**53.32**
STGD 437	***ABCA4***	**NM_000350.2**	**c.4978C>T**	**p.(Pro1660Ser)**	**NC_000001.10:g.94486836G>A**	**het**	**AR**	**P**	**PS1_MODERATE, PP3, PM2, PM5, PM1_SUPPORTING, PP2, PP5_STRONG**	**CM125714**	**-**	**0.0001**	**unknown**	**no**	**negative**	**f**	**53.32**
STGD 438	***ABCA4***	**NM_000350.2**	**c.1442del**	**p.(Phe481Serfs*87)**	**NC_000001.10:g.94543359del**	**het**	**AR**	**P**	**PM2, PVS1, PP5**	**-**	**-**	**-**	**unknown**	**no**	**positive (duplex)**	**f**	**57.29**
STGD 438	***ABCA4***	**NM_000350.2**	**c.5714+5G>A**	**p.(?)**	**NC_000001.10:g.94476351C>T**	**het**	**AR**	**P**	**PP3, PM2, PP5_STRONG, PS4_MODERATE**	**CS982057**	**-**	**0.0003**	**unknown**	**no**	**positive (duplex)**	**f**	**57.29**
STGD 441	***ABCA4***	**NM_000350.2**	**c.1622T>C**	**p.(Leu541Pro)**	**NC_000001.10:g.94528806A>G**	**het**	**AR**	**P**	**PP3, PM2, PP2, PP5_STRONG, PS4_MODERATE**	**CM990022**	**7901**	**0.0003**	**unknown**	**no**	**negative**	**f**	**45.88**
STGD 441	***ABCA4***	**NM_000350.2**	**c.3113C>T**	**p.(Ala1038Val)**	**NC_000001.10:g.94508969G>A**	**het**	**AR**	**P**	**PM2, PM1_SUPPORTING, PP2, PP5_STRONG, PS4_MODERATE**	**CM970006**	**7901**	**0.0024**	**unknown**	**no**	**negative**	**f**	**45.88**
STGD 441	***ABCA4***	**NM_000350.2**	**c.5882G>A**	**p.(Gly1961Glu)**	**NC_000001.10:g.94473807C>T**	**het**	**AR**	**P**	**PP3, PM2, PM5, PP2, PP5_STRONG, PS4_MODERATE**	**CM970016**	**635154**	**0.0047**	**unknown**	**no**	**negative**	**f**	**45.88**
STGD 441	*GUCY2D*	NM_000180.3	c.380C>G	p.(Pro127Arg)	NC_000017.10:g.7906745C>G	het	AR+AD	VUS	PM2, PM5, PP2	-	-	-	unknown	no	negative	f	45.88
STGD 442	***ABCA4***	**NM_000350.2**	**c.5018+2T>C**	**p.(?)**	**NC_000001.10:g.94486794A>G**	**het**	**AR**	**P**	**PM2, PVS1, PP5, PS4**	**CS0911023**	**-**	**-**	**unknown**	**no**	**positive (triplex)**	**f**	**54.08**
STGD 442	***ABCA4***	**NM_000350.2**	**c.5882G>A**	**p.(Gly1961Glu)**	**NC_000001.10:g.94473807C>T**	**het**	**AR**	**P**	**PP3, PM2, PM5, PP2, PP5_STRONG, PS4_MODERATE**	**CM970016**	**635154**	**0.0047**	**unknown**	**no**	**positive (triplex)**	**f**	**54.08**
STGD 442	*CYP4V2*	NM_207352.3	c.1201A>G	p.(Ser401Gly)	NC_000044.11:g.187130129A>G	het	AR	VUS	PM2, PM1_SUPPORTING, PP2, BP4	-	-	0.0001	unknown	no	positive (triplex)	f	54.08
STGD 443	***ABCA4***	**NM_000350.2**	**c.2588G>C**	**p.(Gly863Ala)**	**NC_000001.10:g.94517254C>G**	**het**	**AR**	**P**	**PP3, PM2, PP2, PP5_STRONG, PS4_MODERATE**	**CM970003&CS024003**	**-**	**0.0044**	**unknown**	**no**	**negative**	**f**	**40.50**
STGD 443	***ABCA4***	**NM_000350.2**	**c.4234C>T**	**p.(Gln1412*)**	**NC_000001.10:g.94496571G>A**	**het**	**AR**	**P**	**PM2, PVS1, PP5**	**CM990049**	**-**	**0.0001**	**unknown**	**no**	**negative**	**f**	**40.50**
STGD 443	***ABCA4***	**NM_000350.2**	**c.5603A>T**	**p.(Asn1868Ile)**	**NC_000001.10:g.94476467T>A**	**het**	**AR**	**VUS**	**PP2, PP5, BA1, BS2**	**CM015091**	**-**	**0.0426**	**unknown**	**no**	**negative**	**f**	**40.50**
STGD 443	*RP2*	NM_006915.2	c.58G>C	p.(Glu20Gln)	NC_000023.10:g.46696593G>C	het	XLR+XLD	VUS	PM2	-	-	-	unknown	no	negative	f	40.50
STGD 445	***ABCA4***	**NM_000350.2**	**c.4234C>T**	**p.(Gln1412*)**	**NC_000001.10:g.94496571G>A**	**het**	**AR**	**P**	**PM2, PVS1, PP5**	**CM990049**	**-**	**0.0001**	**unknown**	**no**	**negative**	**f**	**39.08**
STGD 445	***ABCA4***	**NM_000350.2**	**c.4253+43G>A**	**p.(?)**	**NC_000001.10:g.94496509C>T**	**het**	**AR**	**LP**	**PM2, PP5_STRONG, BP7**	**CS193151**	**-**	**0.0047**	**unknown**	**no**	**negative**	**f**	**39.08**
STGD 448	***IMPG1***	**NM_001563.2**	**c.151dup**	**p.(Met51Asnfs*29)**	**NC_000006.11:g.76751765dup**	**het**	**AD**	**P**	**PM2, PVS1, PP5**	**-**	**1275815**	**0.0001**	**unknown**	**no**	**negative**	**m**	**38.50**
STGD 448	*ABCA4*	NM_000350.2	c.181A>G	p.(Met61Val)	NC_000001.10:g.94577115T>C	het	AR	LP	PP3, PM2, PM1, PP2	-	-	-	unknown	no	negative	m	38.50
STGD 448	*ABCA4*	NM_000350.2	c.3703A>G	p.(Asn1235Asp)	NC_000001.10:g.94502811T>C	het	AR	LP	PM2, PP2, PP5_STRONG	CM032811	-	-	unknown	no	negative	m	38.50
STGD 449	***ABCA4***	**NM_000350.2**	**c.1253T>C**	**p.(Phe418Ser)**	**NC_000001.10:g.94544249A>G**	**het**	**AR**	**P**	**PP3, PM2, PP2, PP5_STRONG**	**CM117250**	**-**	**0.0001**	**unknown**	**no**	**negative**	**f**	**38.46**
STGD 449	***ABCA4***	**NM_000350.2**	**c.5196+1137G>A**	**p.(?)**	**NC_000001.10:g.94484001C>T**	**het**	**AR**	**P**	**PM2, PP5_STRONG, BP7, PS4_MODERATE**	**CS1313969**	**438100**	**0.0001**	**unknown**	**no**	**negative**	**f**	**38.46**
STGD 453	***ABCA4***	**NM_000350.2**	**c.5018+2T>C**	**p.(?)**	**NC_000001.10:g.94486794A>G**	**het**	**AR**	**P**	**PM2, PVS1, PP5, PS4**	**CS0911023**	**-**	**-**	**unknown**	**no**	**negative**	**m**	**40.31**
STGD 453	***ABCA4***	**NM_000350.2**	**c.5578C>T**	**p.(Arg1860Trp)**	**NC_000001.10:g.94476824G>A**	**het**	**AR**	**LP**	**PM2, PM1_SUPPORTING, PP2, PP5_STRONG**	**CM1311045**	**1275763**	**0.0001**	**unknown**	**no**	**negative**	**m**	**40.31**
STGD 455	***ABCA4***	**NM_000350.2**	**c.5882G>A**	**p.(Gly1961Glu)**	**NC_000001.10:g.94473807C>T**	**het**	**AR**	**P**	**PP3, PM2, PM5, PP2, PP5_STRONG, PS4_MODERATE**	**CM970016**	**635154**	**0.0047**	**unknown**	**no**	**positive (duplex)**	**f**	**41.96**
STGD 455	***ABCA4***	**NM_000350.2**	**na**	**na**	**NC_000001.10:g.94586187_94587411del**	**het**	**AR**	**LP**	**PM2, PVS1**	**na**	**-**	**na**	**unknown**	**no**	**positive (duplex)**	**f**	**41.96**
STGD 455	*RP11*	NM_178857.5	c.4486A>C	p.(Thr1496Pro)	NC_000008.10:g.10467122T>G	het	AR+AD	VUS	PM2, BP4	-	-	0.0001	unknown	no	positive (duplex)	f	41.96
STGD 455	*RP11*	NM_178857.5	c.5959C>T	p.(Gln1987*)	NC_000008.10:g.10465649G>A	het	AR+AD	P	PM2, PVS1_STRONG, PP5_STRONG	CM120689	-	0.0016	unknown	no	positive (duplex)	f	41.96
STGD 457	***ABCA4***	**NM_000350.2**	**c.5461-10T>C**	**p.(?)**	**NC_000001.10:g.94476951A>G**	**het**	**AR**	**P**	**PM2, PP5_STRONG, BP4, PS4_MODERATE**	**CS057513**	**-**	**0.0002**	**unknown**	**no**	**negative**	**f**	**39.51**
STGD 457	***ABCA4***	**NM_000350.2**	**c.5603A>T**	**p.(Asn1868Ile)**	**NC_000001.10:g.94476467T>A**	**hom**	**AR**	**VUS**	**PP2, PP5, BA1, BS2**	**CM015091**	**-**	**0.0426**	**unknown**	**no**	**negative**	**f**	**39.51**
STGD 457	*FBN2*	NM_001999.3	c.3253A>C	p.(Thr1085Pro)	NC_000005.9:g.127680167T>G	het	AD	VUS	PM2	-	-	0.0001	unknown	no	negative	f	39.51
STGD 457	*GUCY2D*	NM_000180.3	c.2048C>G	p.(Thr683Ser)	NC_000017.10:g.7915859C>G	het	AR+AD	VUS	PM2, PP2	-	-	-	unknown	no	negative	f	39.51
STGD 458	***ABCA4***	**NM_000350.2**	**c.1622T>C**	**p.(Leu541Pro)**	**NC_000001.10:g.94528806A>G**	**het**	**AR**	**P**	**PP3, PM2, PP2, PP5_STRONG, PS4_MODERATE**	**CM990022**	**7901**	**0.0003**	**unknown**	**no**	**negative**	**f**	**37.67**
STGD 458	***ABCA4***	**NM_000350.2**	**c.3113C>T**	**p.(Ala1038Val)**	**NC_000001.10:g.94508969G>A**	**het**	**AR**	**P**	**PM2, PM1_SUPPORTING, PP2, PP5_STRONG, PS4_MODERATE**	**CM970006**	**7901**	**0.0024**	**unknown**	**no**	**negative**	**f**	**37.67**
STGD 458	***ABCA4***	**NM_000350.2**	**c.2401G>A**	**p.(Ala801Thr)**	**NC_000001.10:g.94520853C>T**	**het**	**AR**	**LP**	**PM2, PM1_SUPPORTING, PP2, PP5**	**CM070632**	**631618**	**0.0001**	**unknown**	**no**	**negative**	**f**	**37.67**
STGD 460	***PRPH2***	**NM_000322.4**	**c.318del**	**p.(Leu107Serfs*32)**	**NC_000006.11:g.42689756del**	**het**	**AD**	**P**	**PM2, PVS1, PP5**	**CD2012283**	**-**	**-**	**unknown**	**no**	**negative**	**f**	**39.83**
STGD 461	***ABCA4***	**NM_000350.2**	**c.1622T>C**	**p.(Leu541Pro)**	**NC_000001.10:g.94528806A>G**	**het**	**AR**	**P**	**PP3, PM2, PP2, PP5_STRONG, PS4_MODERATE**	**CM990022**	**7901**	**0.0003**	**unknown**	**no**	**negative**	**f**	**41.97**
STGD 461	***ABCA4***	**NM_000350.2**	**c.3113C>T**	**p.(Ala1038Val)**	**NC_000001.10:g.94508969G>A**	**het**	**AR**	**P**	**PM2, PM1_SUPPORTING, PP2, PP5_STRONG, PS4_MODERATE**	**CM970006**	**7901**	**0.0024**	**unknown**	**no**	**negative**	**f**	**41.97**
STGD 461	***ABCA4***	**NM_000350.2**	**c.5882G>A**	**p.(Gly1961Glu)**	**NC_000001.10:g.94473807C>T**	**het**	**AR**	**P**	**PP3, PM2, PM5, PP2, PP5_STRONG, PS4_MODERATE**	**CM970016**	**635154**	**0.0047**	**unknown**	**no**	**negative**	**f**	**41.97**
STGD 461	*ABCA4*	NM_000350.2	c.3758C>T	p.(Thr1253Met)	NC_000001.10:g.94502756G>A	het	AR	VUS	PM2	CM012036	-	0.0004	unknown	no	negative	f	41.97
STGD 462	***ABCA4***	**NM_000350.2**	**c.5413A>G**	**p.(Asn1805Asp)**	**NC_000001.10:g.94480146T>C**	**het**	**AR**	**LP**	**PP3, PM2, PM1_SUPPORTING, PP2, PP5**	**CM011907**	**-**	**0.00003**	**unknown**	**no**	**negative**	**m**	**44.86**
STGD 462	***ABCA4***	**NM_000350.2**	**c.5603A>T**	**p.(Asn1868Ile)**	**NC_000001.10:g.94476467T>A**	**het**	**AR**	**VUS**	**PP2, PP5, BA1, BS2**	**CM015091**	**-**	**0.0426**	**unknown**	**no**	**negative**	**m**	**44.86**
STGD 462	*ABCA4*	NM_000350.2	c.5691G>T	p.(Gln1897His)	NC_000001.10:g.94476379C>A	het	AR	VUS	PM2	CM213057	-	-	unknown	no	negative	m	44.86
STGD 464	***ABCA4***	**NM_000350.2**	**c.3386G>T**	**p.(Arg1129Leu)**	**NC_000001.10:g.94506901C>A**	**het**	**AR**	**P**	**PP3, PM2, PM5, PM1, PP2, PP5_STRONG**	**CM970009**	**-**	**0.0003**	**unknown**	**no**	**negative**	**f**	**40.15**
STGD 464	***ABCA4***	**NM_000350.2**	**c.5603A>T**	**p.(Asn1868Ile)**	**NC_000001.10:g.94476467T>A**	**het**	**AR**	**VUS**	**PP2, PP5, BA1, BS2**	**CM015091**	**-**	**0.0426**	**unknown**	**no**	**negative**	**f**	**40.15**
STGD 465	***ABCA4***	**NM_000350.2**	**c.1622T>C**	**p.(Leu541Pro)**	**NC_000001.10:g.94528806A>G**	**het**	**AR**	**P**	**PP3, PM2, PP2, PP5_STRONG, PS4_MODERATE**	**CM990022**	**7901**	**0.0003**	**unknown**	**no**	**unknown**	**m**	**38.31**
STGD 465	***ABCA4***	**NM_000350.2**	**c.3113C>T**	**p.(Ala1038Val)**	**NC_000001.10:g.94508969G>A**	**het**	**AR**	**P**	**PM2, PM1_SUPPORTING, PP2, PP5_STRONG, PS4_MODERATE**	**CM970006**	**7901**	**0.0024**	**unknown**	**no**	**unknown**	**m**	**38.31**
STGD 465	***ABCA4***	**NM_000350.2**	**c.5313-2A>G**	**p.(?)**	**NC_000001.10:g.94480248T>C**	**het**	**AR**	**P**	**PM2, PVS1, PP5**	**-**	**-**	**0.0001**	**unknown**	**no**	**unknown**	**m**	**38.31**
STGD 465	***CACNA1F***	**NM_005183.2**	**c.3167dup**	**p.(Leu1057Glyfs*10)**	**NC_000023.10:g.49072944dup**	**hemi**	**XLR**	**LP**	**PM2, PVS1**	**CI983046**	**-**	**-**	**unknown**	**no**	**unknown**	**m**	**38.31**
STGD 465	*CNGB3*	NM_019098.4	c.890_896del	p.(Ser297Tyrfs*9)	NC_000008.10:g.87666247_87666253del	het	AR	LP	PM2, PVS1	-	-	-	unknown	no	unknown	m	38.31
USH I 3	***MYO7A***	**NM_000260.3**	**c.494C>T**	**p.(Thr165Met)**	**NC_000011.9:g.76867729C>T**	**het**	**AR**	**P**	**PP3, PM2, PP5_STRONG**	**CM050717**	**-**	**0.0001**	**unknown**	**single genes**	**negative**	**f**	**39.70**
USH I 3	***MYO7A***	**NM_000260.3**	**c.849+5G>A**	**p.(?)**	**NC_000011.9:g.76868443G>A**	**het**	**AR**	**LP**	**PP3, PM2, PP5_STRONG**	**CS1713761**	**-**	**0.0001**	**unknown**	**single genes**	**negative**	**f**	**39.70**
USH I 3	*MYO7A*	NM_000260.3	c.2210A>G	p.(Glu737Gly)	NC_000011.9:g.76888617A>G	het	AR	VUS	PP3, PM2	CM1619349	-	0.0001	unknown	single genes	negative	f	39.70
USH I 21	***PCDH15***	**NM_033056.3**	**c.4128dup**	**p.(Leu1378Valfs*26)**	**NC_000010.10:g.55591150dup**	**het**	**AR**	**P**	**PM2, PVS1, PP5**	**CI1619505**	**1073474**	**-**	**unknown**	**no**	**positive (multiplex)**	**m**	**40.09**
USH I 21	***PCDH15***	**NM_033056.3**	**c.986-5151_993del**	**p.(?)**	**NC_000010.10:g.55973806_55978964del**	**het**	**AR**	**LP**	**PM2, PVS1**	**na**	**-**	**na**	**unknown**	**no**	**positive (multiplex)**	**m**	**40.09**
USH I 53	***CLRN1***	**NM_174878.2**	**c.5287>G**	**p.(Tyr176*)**	**NC_000003.11:g.150645894A>C**	**hom**	**AR**	**P**	**PM2, PVS1_STRONG, PP5_STRONG**	**CM013798**	**4392**	**0.000004**	**unknown**	**no**	**unknown**	**m**	**51.70**
USH I 63	***CDH23***	**NM_022124.5**	**c.6050-9G>A**	**p.(?)**	**NC_000010.10:g.73550880G>A**	**hom**	**AR**	**LP**	**PM2, PP3, PS3**	**CS020531**	**46001**	**0.00005**	**unknown**	**no**	**positive (duplex)**	**m**	**50.47**
USH I 73	***MYO7A***	**NM_000260.3**	**c.494C>T**	**p.(Thr165Met)**	**NC_000011.9:g.76867729C>T**	**het**	**AR**	**P**	**PP3, PM2, PP5_STRONG**	**CM050717**	**43264**	**0.0001**	**unknown**	**no**	**negative**	**f**	**45.42**
USH I 73	***MYO7A***	**NM_000260.3**	**c.5573T>C**	**p.(Leu1858Pro)**	**NC_000011.9:g.76916599T>C**	**het**	**AR**	**P**	**PP3, PM2, PP5_STRONG**

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XRP 188	***RPGR***	**NM_001034853.1**	**c.379del**	**p.(Arg127Glufs*6)**	**NC_000023.10:g.38178174del**	**hemi**	**XLR+XLD**	**P**	**PM2, PVS1, PP5**	**-**	**-**	**-**	**unknown**	**single genes**	**positive (duplex)**	**m**	**53.95**
XRP 188	*PRPF31*	NM_015629.3	c.793A>G	p.(Thr265Ala)	NC_000019.9:g.54627973A>G	het	AD	VUS	PM2, PP2	-	-	-	unknown	single genes	positive (duplex)	m	53.95
XRP 198	***RPGR***	**NM_001034853.1**	**c.644G>T**	**p.(Gly215Val)**	**NC_000023.10:g.38170002C>A**	**het**	**XLR+XLD**	**LP**	**PP3, PM2, PP5_STRONG**	**CM961259**	**-**	**-**	**unknown**	**no**	**positive (multiplex)**	**f**	**57.86**
XRP 199	***RP2***	**NM_006915.2**	**c.358C>T**	**p.(Arg120*)**	**NC_000023.10:g.46713166C>T**	**het**	**XLR+XLD**	**P**	**PM2, PVS1, PP5, PS4**	**CM991106**	**-**	**-**	**unknown**	**no**	**negative**	**f**	**48.94**
XRP 199	*EYS*	NM_001142800.1	c.5086G>T	p.(Glu1696*)	NC_000006.11:g.65300674C>A	het	AR	LP	PM2, PVS1	-	-	-	unknown	no	negative	f	48.94
XRP 203	***RPGR***	**NM_001034853.1**	**c.3049G>T**	**p.(Glu1017*)**	**NC_000023.10:g.38145203C>A**	**hemi**	**XLR+XLD**	**P**	**PM2, PVS1, PP5**	**CM2036892**	**-**	**-**	**unknown**	**no**	**negative**	**m**	**47.66**
XRP 224	***RP2***	**NM_006915.2**	**c.769-3C>A**	**p.(?)**	**NC_000023.10:g.46719420C>A**	**het**	**XLR+XLD**	**LP**	**PP3, PM2, PP5_STRONG**	**CS083269**	**-**	**-**	**unknown**	**no**	**positive (duplex)**	**f**	**52.03**
XRP 226	***RPGR***	**NM_001034853.1**	**c.1281_1282del**	**p.(Leu428Thrfs*24)**	**NC_000023.10:g.38156671_38156672del**	**hemi**	**XLR+XLD**	**P**	**PM2, PVS1, PP5**	**-**	**-**	**-**	**unknown**	**single genes**	**positive (multiplex)**	**m**	**50.76**
XRP 228	***RPGR***	**NM_001034853.1**	**c.2476_2477del**	**p.(Arg826Glyfs*8)**	**NC_000023.10:g.38145780_38145781del**	**hemi**	**XLR+XLD**	**P**	**PM2, PVS1, PP5**	**CD022919**	**-**	**-**	**unknown**	**no**	**negative**	**m**	**54.00**
XRP 230	***RPGR***	**NM_001034853.1**	**c.2236_2237del**	**p.(Glu746Argfs*23)**	**NC_000023.10:g.38146018_38146019del**	**het**	**XLR+XLD**	**LP**	**PM2, PVS1_STRONG**	**CD2026823, CD004113**	**438142**	**-**	**unknown**	**no**	**positive (duplex)**	**f**	**54.57**
XRP 231	***RPGR***	**NM_001034853.1**	**c.2689G>T**	**p.(Glu897*)**	**NC_000023.10:g.38145563C>A**	**het**	**XLR+XLD**	**P**	**PM2, PVS1, PP5**	**CM140498**	**-**	**-**	**unknown**	**no**	**positive (duplex)**	**f**	**47.41**
XRP 232	***RPGR***	**NM_001034853.1**	**c.779-5T>G**	**p.(?)**	**NC_000023.10:g.38164048A>C**	**hemi**	**XLR+XLD**	**LP**	**PM2, PP5_STRONG, BP4**	**CS083271**	**-**	**-**	**unknown**	**no**	**positive (duplex)**	**m**	**51.52**
XRP 233	***RP2***	**NM_006915.2**	**c.352C>T**	**p.(Arg118Cys)**	**NC_000023.10:g.46713160C>T**	**hemi**	**XLR+XLD**	**P**	**PP3, PM2, PM5, PM1_SUPPORTING, PP5_STRONG, PS4_MODERATE**	**CM033008**	**-**	**-**	**unknown**	**no**	**positive (duplex)**	**m**	**50.99**
XRP 234	***RPGR***	**NM_001034853.1**	**c.2780_2782delinsGAGGAGGAAGGAGAGGAAT**	**p.(Glu927Glyfs*157)**	**NC_000023.10:g.38145470_38145472delinsATTCCTCTCCTTCCTCCTC**	**hemi**	**XLD+XLR**	**LP**	**PVS1, PM2**	**-**	**-**	**-**	**unknown**	**no**	**positive (duplex)**	**m**	**48.43**
XRP 234	*RP1L1*	NM_178857.5	c.5821C>T	p.(Gln1941*)	NC_000008.10:g.10465787G>A	het	AD+AR	LP	PM2, PVS1_STRONG, PP5	CM2035439	-	0.0005	unknown	no	positive (duplex)	m	48.43
XRP 235	***RPGR***	**NM_001034853.1**	**c.1573-12A>G**	**p.(?)**	**NC_000023.10:g.38147306T>C**	**hemi**	**XLR+XLD**	**LP**	**PM2, PS3**	**-**	**-**	**-**	**unknown**	**no**	**negative**	**m**	**47.55**
XRP 235	*IMPDH1*	NM_000883.3	c.590A>G	p.(Gln197Arg)	NC_000007.13:g.128040583T>C	het	AD	VUS	PM2	-	-	0.0001	unknown	no	negative	m	47.55
XRP 236	***RPGR***	**NM_001034853.1**	**c.2323_2324del**	**p.(Arg775Glufs*59)**	**NC_000023.10:g.38145933_38145934del**	**hemi**	**XLR+XLD**	**LP**	**PM2, PVS1_STRONG**	**CD022917**	**-**	**-**	**unknown**	**no**	**negative**	**m**	**41.51**
XRP 237	***RP2***	**NM_006915.2**	**c.358C>T**	**p.(Arg120*)**	**NC_000023.10:g.46713166C>T**	**het**	**XLR+XLD**	**P**	**PM2, PVS1, PP5, PS4**	**CM991106**	**-**	**-**	**unknown**	**no**	**positive (triplex)**	**f**	**40.29**
XRP 238	***RPGR***	**NM_001034853.1**	**c.2501del**	**p.(Glu834Glyfs*255)**	**NC_000023.10:g.38145751del**	**hemi**	**XLR+XLD**	**P**	**PM2, PVS1, PP5**	**CD033259**	**-**	**-**	**unknown**	**no**	**positive (duplex)**	**m**	**53.36**
XRP 239	***RP2***	**NM_006915.2**	**na**	**na**	**NC_000023.10:g.(46737528_46737831)_46745341delins[NC_000002.11:g.(158406216_158406544)_158695305;GTTG]**	**hemi**	**XLR+XLD**	**P**	**PM2, PVS1, PP5**	**na**	**-**	**na**	**unknown**	**no**	**positive (multiplex)**	**m**	**44.35**
XRP 240	***RP2***	**NM_006915.2**	**na**	**na**	**NC_000023.10:g.46694894_46708019del**	**hemi**	**XLR+XLD**	**P**	**PM2, PVS1, PP5**	**na**	**-**	**na**	**unknown**	**no**	**negative**	**m**	**42.44**
XRP 241	***RPGR***	**NM_001034853.1**	**c.2763_2764del**	**p.(Glu922Glyfs*156)**	**NC_000023.10:g.38145490_38145491del**	**hemi**	**XLR+XLD**	**P**	**PM2, PVS1, PP5**	**CD004120**	**-**	**-**	**unknown**	**no**	**negative**	**m**	**49.10**
XRP 241	*GUCY2D*	NM_000180.3	c.2302C>T	p.(Arg768Trp)	NC_000017.10:g.7917236C>T	het	AR+AD	P	PS1_MODERATE, PP3, PM2, PM5, PP2, PP5_STRONG	CM004764	-	0.0002	unknown	no	negative	m	49.10
XRP 242	***RPGR***	**NM_001034853.1**	**c.2548G>T**	**p.(Glu850*)**	**NC_000023.10:g.38145704C>A**	**hemi**	**XLR+XLD**	**P**	**PM2, PVS1, PP5**	**-**	**-**	**-**	**unknown**	**MIP**	**positive (duplex)**	**m**	**52.85**
XRP 243	***RPGR***	**NM_001034853.1**	**c.1216_1217del**	**p.(Leu406Ilefs*46)**	**NC_000023.10:g.38158239_38158240del**	**hemi**	**XLR+XLD**	**P**	**PM2, PVS1, PP5**	**CD033232**	**975132**	**-**	**unknown**	**no**	**positive (duplex)**	**m**	**55.15**
XRP 244	***RPGR***	**NM_001034853.1**	**c.1932_1933insC**	**p.(Gly645Argfs*8)**	**NC_000023.10:g.38146319_38146320insG**	**het**	**XLR+XLD**	**LP**	**PM2, PVS1**	**-**	**1184953**	**-**	**unknown**	**no**	**positive (duplex)**	**f**	**58.98**
XRP 245	***RPGR***	**NM_001034853.1**	**c.29-1G>T**	**p.(?)**	**NC_000023.10:g.38182778C>A**	**hemi**	**XLR+XLD**	**P**	**PM2, PVS1_STRONG, PP5_STRONG**	**CS1825645**	**813233**	**-**	**unknown**	**no**	**negative**	**m**	**55.07**
XRP 246	***RP2***	**NM_006915.2**	**c.838del**	**p.(Val280Phefs*13)**	**NC_000023.10:g.46719492del**	**hemi**	**XLR+XLD**	**P**	**PM2, PVS1, PP5**	**-**	**1184894**	**-**	**unknown**	**no**	**unknown**	**m**	**40.43**
XRP 248	***RPGR***	**NM_001034853.1**	**na**	**na**	**NC_000023.10:g.38184601_38194323delinsTACACAG**	**het**	**XLR+XLD**	**P**	**PM2, PVS1, PP5**	**na**	**-**	**na**	**unknown**	**no**	**positive (multiplex)**	**f**	**42.49**
XRP 248	*RP1*	NM_006269.1	c.652G>A	p.(Ala218Thr)	NC_000008.10:g.55534713G>A	het	AD	VUS	PM2, BS2_SUPPORTING	CM014380	-	0.0003	unknown	no	positive (multiplex)	f	42.49
XRP 252	***RPGR***	**NM_001034853.1**	**c.2218G>T**	**p.(Glu740*)**	**NC_000023.10:g.38146034C>A**	**hemi**	**XLR+XLD**	**P**	**PM2, PVS1, PP5**	**CM020791**	**1275779**	**-**	**unknown**	**no**	**positive (duplex)**	**m**	**41.84**
XRP 255	***RP2***	**NM_006915.2**	**c.14_16del**	**p.(Phe5del)**	**NC_000023.10:g.46696549_46696551del**	**hemi**	**XLR+XLD**	**P**	**PM2, PM4, PP5_STRONG, PS4_MODERATE**	**CD083343**	**-**	**-**	**maternal**	**no**	**negative**	**m**	**42.52**
XRP 258	***RPGR***	**NM_001034853.1**	**c.1245+224_1414+653del**	**p.(?)**	**NC_000023.10:g.38155887_38157988del**	**hemi**	**XLR+XLD**	**P**	**PM2, PVS1, PP5**	**na**	**-**	**na**	**unknown**	**no**	**positive (multiplex)**	**f**	**41.07**
Possibly solved cases																			
ACHM 497	*CNGA3*	NM_001298.2	c.1641C>A	p.(Phe547Leu)	NC_000002.11:g.99013274C>A	het	AR	P	PS1_MODERATE, PP3, PM2, PM5_SUPPORTING, PM1, PP2, PP5_STRONG	CM980380	-	0.0002	unknown	single genes	negative	f	51.30
ACHM 497	*CNGA3*	NM_001298.2	c.479T>G	p.(Val160Gly)	NC_000002.11:g.99006150T>G	het	AR	VUS	PP3, PM2, PM1_SUPPORTING, PP2	-	-	-	unknown	single genes	negative	f	51.30
ACHM 1192	*PDE6C*	NM_006204.3	c.752T>C	p.(Phe251Ser)	NC_000010.10:g.95381717T>C	hom	AR	VUS	PM2	-	-	-	unknown	no	positive (multiplex)	f	45.32
ACHM 1207	*PDE6C*	NM_006204.3	c.1071+5G>A	p.(?)	NC_000010.10:g.95386633G>A	hom	AR	VUS	PP3, PM2	-	-	-	unknown	no	negative	m	55.31
ACHM 1247	*CNGB3*	NM_019098.4	c.1148del	p.(Thr383Ilefs*13)	NC_000008.10:g.87656009del	het	AR	P	PM2, PVS1, PP5, PS4_MODERATE	CD001927	-	0.0020	unknown	no	negative	m	56.32
ACHM 1247	*CNGB3*	NM_019098.4	c.1179-11T>G	p.(?)	NC_000008.10:g.87645132A>C	het	AR	VUS	PM2, BP7	-	-	-	unknown	no	negative	m	56.32
ACHM 1284	*RP1L1*	NM_178857.5	c.2882T>C	p.(Leu961Pro)	NC_000008.10:g.10468726A>G	hom	AD+AR	VUS	PM2	-	-	-	unknown	no	positive (duplex)	m	49.36
ACHM 1284	*RPGR*	NM_001034853.1	c.2633_2664delinsAAGAAGGG	p.(Gly878_Glu888delinsGluGluGly)	NC_000023.10:g.38145588_38145619delinsCCCTTCTT	hemi	XLR+XLD	VUS	PM2	-	-	-	unknown	no	positive (duplex)	m	49.36
ADRP 449	*LAMA1*	NM_005559.3	c.176G>A	p.(Arg59Gln)	NC_000018.9:g.7080342C>T	het	AR	VUS	PM2, BP4	-	-	0.00002	unknown	no	positive (triplex)	m	48.20
ADRP 449	*LAMA1*	NM_005559.3	c.2293A>G	p.(Thr765Ala)	NC_000018.9:g.7026087T>C	het	AR	VUS	PM2, BP4	-	-	-	unknown	no	positive (triplex)	m	48.20
ADRP 462	*SNRNP200*	NM_014014.4	c.4171A>G	p.(Met1391Val)	NC_000002.11:g.96950317T>C	het	AD	VUS	PM2, PP2, BP4	-	-	-	unknown	no	positive (triplex)	f	46.41
ADRP 466	*PRPF8*	NM_006445.3	c.6838A>G	p.(Asn2280Asp)	NC_000017.10:g.1554417T>C	het	AD	VUS	PP3, PM2, PP2	-	-	-	unknown	no	positive (multiplex)	m	46.71
ADRP 503	*IMPDH1*	NM_000883.3	c.730A>C	p.(Thr244Pro)	NC_000007.13:g.128040443T>G	het	AD	VUS	PP3, PM2	-	-	0.0001	unknown	no	positive (duplex)	f	55.28
ADRP 507	*PRPF3*	NM_004698.2	c.1496A>C	p.(His499Pro)	NC_000001.10:g.150316707A>C	het	AD	VUS	PP3, PM2, PP2, PP5	CM2036738	-	-	unknown	no	positive (multiplex)	m	56.32
ADRP 511-1	*PRPH2*	NM_000322.4	c.751_752delinsCC	p.(Arg251Pro)	NC_000006.11:g.42672179_42672180delinsGG	het	AD	VUS	PM2, PM1, PP2	-	-	-	unknown	no	positive (multiplex)	f	62.68
ADRP 511-1	*CERKL*	NM_201548.4	c.769C>T	p.(Arg257*)	NC_000002.11:g.182423344G>A	het	AR	P	PM2, PVS1, PP5, PS4	CM040509	-	0.0003	unknown	no	positive (multiplex)	f	62.68
ADRP 511-2	*PRPH2*	NM_000322.4	c.751_752delinsCC	p.(Arg251Pro)	NC_000006.11:g.42672179_42672180delinsGG	het	AD	VUS	PM2, PM1, PP2	-	-	-	unknown	no	positive (multiplex)	f	62.14
ARRP 186	*RDH12*	NM_152443.2	c.481C>T	p.(Arg161Trp)	NC_000014.8:g.68193730C>T	het	AR	LP	PM2, PP2, PP5_STRONG	CM118733	-	0.0001	unknown	no	positive (duplex)	f	38.32
ARRP 186	*RDH12*	NM_152443.2	c.632C>T	p.(Thr211Ile)	NC_000014.8:g.68193881C>T	het	AR	VUS	PP3, PM2, PM1_SUPPORTING, PP2	-	-	-	unknown	no	positive (duplex)	f	38.32
ARRP 186	*SEMA4A*	NM_022367.3	c.149G>A	p.(Arg50His)	NC_000001.10:g.156126214G>A	het	AD	VUS	PM2, BP4	-	-	0.0001	unknown	no	positive (duplex)	f	38.32
ARRP 285	*SDCCAG8*	NM_006642.3	c.1946_1949del	p.(Cys6495efs*10)	NC_000001.10:g.243589821_243589824del	het	AR	LP	PM2, PVS1	CD106174	-	-	unknown	Panel	positive (duplex)	f	41.04
ARRP 285	*SDCCAG8*	NM_006642.3	c.740+267C>T	p.(?)	NC_000001.10:g.243468346C>T	het	AR	VUS	PM2	-	-	-	unknown	Panel	positive (duplex)	f	41.04
ARRP 331-1	*CDH23*	NM_022124.5	c.2569G>A	p.(Val857Ile)	NC_000010.10:g.73461950G>A	het	AR	VUS	PM2	-	-	0.0001	unknown	Panel	positive (duplex)	f	59.50
ARRP 331-1	*CDH23*	NM_022124.5	c.6856G>A	p.(Val2286Ile)	NC_000010.10:g.73558137G>A	het	AR	VUS	PM2	-	-	0.0001	unknown	Panel	positive (duplex)	f	59.50
ARRP 331-1	*TYR*	NM_000372.4	c.1205G>A	p.(Arg402Gln)	NC_000011.9:g.89017961G>A	het	AR	VUS	PM5_SUPPORTING, PP2, PP5, BP6	CM971555	-	0.1769	unknown	Panel	positive (duplex)	f	59.50
ARRP 331-1	*TYR*	NM_000372.4	c.902C>T	p.(Pro301Leu)	NC_000011.9:g.88924452C>T	het	AR	LP	PP3, PM2, PM1, PP2, PP5	CM156816	-	-	unknown	Panel	positive (duplex)	f	59.50
ARRP 331-2	*CDH23*	NM_022124.5	c.2569G>A	p.(Val857Ile)	NC_000010.10:g.73461950G>A	het	AR	VUS	PM2	-	-	0.0001	unknown	Panel	positive (duplex)	m	54.54
ARRP 331-2	*CDH23*	NM_022124.5	c.6856G>A	p.(Val2286Ile)	NC_000010.10:g.73558137G>A	het	AR	VUS	PM2	-	-	0.0001	unknown	Panel	positive (duplex)	m	54.54
ARRP 331-2	*TYR*	NM_000372.4	c.1205G>A	p.(Arg402Gln)	NC_000011.9:g.89017961G>A	het	AR	VUS	PM5_SUPPORTING, PP2, PP5, BP6	CM971555	-	0.1769	unknown	Panel	positive (duplex)	m	54.54
ARRP 331-2	*TYR*	NM_000372.4	c.902C>T	p.(Pro301Leu)	NC_000011.9:g.88924452C>T	het	AR	LP	PP3, PM2, PM1, PP2, PP5	CM156816	-	-	unknown	Panel	positive (duplex)	m	54.54
ARRP 331-2	*PCDH15*	NM_033056.3	c.4644_4689dup	p.(Arg1564Cysfs*13)	NC_000010.10:g.55582797_55582842dup	het	AR	P	PM2, PVS1_STRONG, PP5_STRONG	-	-	-	unknown	Panel	positive (duplex)	m	54.54
ARRP 353	*LAMA1*	NM_005559.3	c.6053C>G	p.(Ala2018Gly)	NC_000018.9:g.6978332G>C	het	AR	VUS	PM2	-	-	-	unknown	no	positive (duplex)	f	50.29
ARRP 353	*LAMA1*	NM_005559.3	c.6968C>T	p.(Pro2323Leu)	NC_000018.9:g.6966228G>A	het	AR	VUS	PM2	-	-	0.000046	unknown	no	positive (duplex)	f	50.29
ARRP 371	*RPGR*	NM_001034853.1	c.2594_2596del	p.(Gly865del)	NC_000023.10:g.38145658_38145660del	het	XLR+XLD	VUS	PM2	CD2035521	-	-	unknown	Panel	negative	f	34.16
ARRP 417	*BBS1*	NM_024649.4	c.526C>T	p.(Gln176*)	NC_000011.9:g.66283339C>T	het	AR	LP	PM2, PVS1	-	-	-	unknown	no	negative	f	30.89
ARRP 417	*TULP1*	NM_003322.3	c.1507_1521del	p.(Val503_Gly507del)	NC_000006.11:g.35466213_35466227del	hom	AR	VUS	PM2, PM4	-	-	-	unknown	no	negative	f	30.89
ARRP 426	*PITPNM3*	NM_031220.3	c.16C>T	p.(Arg6Cys)	NC_000017.10:g.6459711G>A	het	AD	VUS	PM2, BP4	-	-	0.00002	unknown	no	positive (duplex)	f	51.54
ARRP 427	*SNRNP200*	NM_014014.4	c.1082A>G	p.(Tyr361Cys)	NC_000002.11:g.96964059T>C	het	AD	VUS	PM2, PP2	-	-	0.000004	unknown	no	positive (duplex)	m	50.28
ARRP 427	*PRPF8*	NM_006445.3	c.37C>G	p.(Pro13Ala)	NC_000017.10:g.1587829G>C	het	AD	VUS	PM2, PP2	CM2116546	-	0.0001	unknown	no	positive (duplex)	m	50.28
ARRP 427	*USH2A*	NM_206933.2	c.2276G>T	p.(Cys759Phe)	NC_000001.10:g.216420460C>A	het	AR	P	PP3, PM2, PM1_SUPPORTING, PP5_STRONG, PS4_MODERATE	CM001372	2356	0.00097	unknown	no	positive (duplex)	m	50.28
ARRP 428	*NR2E3*	NM_014249.3	c.1171_1172del	p.(Phe391Profs*15)	NC_000015.9:g.72109963_72109964del	het	AR+AD	LP	PM2, PVS1	CD2119618	-	-	unknown	no	positive (duplex)	m	52.54
ARRP 428	*NR2E3*	NM_014249.3	c.151G>A	p.(Gly51Arg)	NC_000015.9:g.72103855G>A	het	AR+AD	VUS	PM2, PM1_SUPPORTING, PP5	CM134234	-	0.000019	unknown	no	positive (duplex)	m	52.54
ARRP 444	*EYS*	NM_001142800.1	c.2382-5T>A	p.(?)	NC_000006.11:g.65622641A>T	hom	AR	VUS	PM2	-	-	-	unknown	no	negative	m	56.06
ARRP 444	*REEP6*	NM_138393.1	c.469G>A	p.(Asp157Asn)	NC_000019.9:g.14964046G>A	hom	AR	VUS	PM2, BP4	-	-	0.0011	unknown	no	negative	m	56.06
ARRP 444	*RPGR*	NM_001034853.1	c.2594_2596del	p.(Gly865del)	NC_000023.10:g.38145658_38145660del	hemi	XLR+XLD	VUS	PM2	CD2035521	-	-	unknown	no	negative	m	56.06
ARRP 445	*EYS*	NM_001142800.1	c.490C>T	p.(Arg164*)	NC_000006.11:g.66204814G>A	het	AR	P	PM2, PVS1, PP5	CM1211748	-	0.0001	unknown	no	negative	m	64.97
ARRP 445	*EYS*	NM_001142800.1	c.5957T>A	p.(Leu1986Gln)	NC_000006.11:g.65098704A>T	het	AR	VUS	PM2	-	-	-	unknown	no	negative	m	64.97
ARRP 445	*EYS*	NM_001142800.1	c.8861T>A	p.(Phe2954Tyr)	NC_000006.11:g.64431066A>T	het	AR	VUS	PM2	-	-	0.0001	unknown	no	negative	m	64.97
ARRP 451	*PDE6A*	NM_000440.2	c.816T>A	p.(Cys272*)	NC_000005.9:g.149310633A>T	hom	AR	P	PM2, PVS1, PP5	-	-	-	unknown	no	negative	m	57.36
ARRP 451	*PDE6A*	NM_000440.2	c.858+5G>T	p.(?)	NC_000005.9:g.149310586C>A	hom	AR	VUS	PP3, PM2	-	-	-	unknown	no	negative	m	57.36
ARRP 452	*RDH12*	NM_152443.2	c.641T>C	p.(Leu214Pro)	NC_000014.8:g.68193890T>C	hom	AR	VUS	PP3, PM2, PM1_SUPPORTING, PP2	CM1817940	-	-	unknown	no	positive (triplex)	m	55.36
ARRP 459	*EYS*	NM_001142800.1	c.9376_9379del	p.(Ile3127Asnfs*2)	NC_000006.11:g.64430550_64430553del	het	AR	LP	PM2, PVS1	CD199508	-	0.0001	unknown	no	positive (duplex)	m	67.28
ARRP 459	*EYS*	NM_001142800.1	c.9410G>A	p.(Gly3137Glu)	NC_000006.11:g.64430517C>T	het	AR	VUS	PM2	-	-	0.0001	unknown	no	positive (duplex)	m	67.28
ARRP 467	*EYS*	NM_001142800.1	c.4789_4801del	p.(Tyr1597Glufs*17)	NC_000006.11:g.65300962_65300974del	het	AR	LP	PM2, PVS1	-	-	-	unknown	no	negative	f	51.48
ARRP 467	*EYS*	NM_001142800.1	c.7811G>A	p.(Arg2604His)	NC_000006.11:g.64487986C>T	het	AR	VUS	PM2, PM1_SUPPORTING	CM180871	-	0.0001	unknown	no	negative	f	51.48
ARRP 467	*ROM1*	NM_000327.3	c.64T>G	p.(Trp22Gly)	NC_000011.9:g.62380817T>G	het	AR+AD	VUS	PP3, PM2	-	-	0.0001	unknown	no	negative	f	51.48
ARRP 468-1	*USH2A*	NM_206933.2	c.13238C>A	p.(Pro4413His)	NC_000001.10:g.215848015G>T	het	AR	VUS	PM2, PM1_SUPPORTING	-	-	-	unknown	no	positive (duplex)	f	53.43
ARRP 468-1	*USH2A*	NM_206933.2	c.8426G>A	p.(Ser2809Asn)	NC_000001.10:g.216052238C>T	het	AR	VUS	PM2	-	-	-	unknown	no	positive (duplex)	f	53.43
ARRP 468-1	*ABCC6*	NM_001171.5	c.1132C>T	p.(Gln378*)	NC_000016.9:g.16295902G>A	het	AR	P	PM2, PVS1, PP5	CM012720	-	0.0002	unknown	no	positive (duplex)	f	53.43
ARRP 468-1	*MEN1*	NM_130799.2	c.1270dup	p.(Glu424Glyfs*25)	NC_000011.9:g.64572588dup	het	AD	LP	PM2, PVS1	CI021455	-	-	unknown	no	positive (duplex)	f	53.43
ARRP 468-2	*USH2A*	NM_206933.2	c.13238C>A	p.(Pro4413His)	NC_000001.10:g.215848015G>T	het	AR	VUS	PM2, PM1_SUPPORTING	-	-	-	unknown	no	positive (duplex)	f	57.34
ARRP 468-2	*USH2A*	NM_206933.2	c.8426G>A	p.(Ser2809Asn)	NC_000001.10:g.216052238C>T	het	AR	VUS	PM2	-	-	-	unknown	no	positive (duplex)	f	57.34
ARRP 468-2	*ABCC6*	NM_001171.5	c.1132C>T	p.(Gln378*)	NC_000016.9:g.16295902G>A	het	AR	P	PM2, PVS1, PP5	CM012720	-	0.0002	unknown	no	positive (duplex)	f	57.34
ARRP 468-2	*MEN1*	NM_130799.2	c.1270dup	p.(Glu424Glyfs*25)	NC_000011.9:g.64572588dup	het	AD	LP	PM2, PVS1	CI021455	-	-	unknown	no	positive (duplex)	f	57.34
ARRP 470	*PDE6B*	NM_000283.3	c.582C>G	p.(Asn194Lys)	NC_000004.11:g.628579C>G	hom	AR	VUS	PM2	-	-	-	unknown	no	negative	m	57.54
ARRP 474	*USH2A*	NM_206933.2	c.12463C>T	p.(Pro4155Ser)	NC_000001.10:g.215848790G>A	hom	AR	VUS	PP3, PM2	-	-	-	unknown	no	negative	m	54.96
ARRP 474	*EYS*	NM_001142800.1	c.5928-2A>G	p.(?)	NC_000006.11:g.65098735T>C	het	AR	P	PM2, PVS1, PP5, PS4_MODERATE	CS119934	-	0.0001	unknown	no	negative	m	54.96
ARRP 489	*USH2A*	NM_206933.2	c.11816_11822dup	p.(Val3942Ilefs*7)	NC_000001.10:g.215901618_215901624dup	het	AR	LP	PM2, PVS1	-	-	-	unknown	no	positive (duplex)	f	41.33
ARRP 489	*USH2A*	NM_206933.2	c.12823T>A	p.(Ser4275Thr)	NC_000001.10:g.215848430A>T	het	AR	VUS	PM2	CM174550	-	0.0006	unknown	no	positive (duplex)	f	41.33
ARRP 489	*USH2A*	NM_206933.2	c.2072G>A	p.(Cys691Tyr)	NC_000001.10:g.216424340C>T	het	AR	VUS	PM2	CM095918	-	-	unknown	no	positive (duplex)	f	41.33
ARRP 489	*MYO7A*	NM_000260.3	c.494C>T	p.(Thr165Met)	NC_000011.9:g.76867729C>T	het	AR	P	PP3, PM2, PP5_STRONG	CM050717	-	0.00003	unknown	no	positive (duplex)	f	41.33
ARRP 495	*EYS*	NM_001142800.1	c.2854del	p.(Cys952Valfs*16)	NC_000006.11:g.65559734del	het	AR	LP	PM2, PVS1	-	-	-	unknown	no	positive (duplex)	m	36.01
ARRP 495	*EYS*	NM_001142800.1	c.9053T>G	p.(Leu3018Trp)	NC_000006.11:g.64430874A>C	het	AR	VUS	PM2	-	-	-	unknown	no	positive (duplex)	m	36.01
BBS 43	*BBS5*	NM_152384.3	na	na	NC_000002.11:g.170343574_170343578ins(400_500)	hom	AR	VUS	PM2	na	-	na	unknown	no	negative	f	52.46
BBS 73	*BBS10*	NM_024685.3	c.271dup	p.(Cys91Leufs*5)	NC_000012.11:g.76741496dup	het	AR	P	PM2, PVS1_STRONG, PP5, PS4	CI064666	-	0.0006	unknown	no	negative	m	47.15
BBS 73	*BBS10*	NM_024685.3	c.1013_1015del	p.(Glu338del)	NC_000012.11:g.76740755_76740757del	het	AR	VUS	PM2, PM4, PM1_SUPPORTING	CD1411589	-	0.0001	unknown	no	negative	m	47.15
BVMD 35	*GUCY2D*	NM_000180.3	c.755G>C	p.(Gly252Ala)	NC_000017.10:g.7907203G>C	het	AD+AR	VUS	PM2, PM1_SUPPORTING, PP2	-	-	0.0001	unknown	single genes	unknown	f	36.12
BVMD 35	*CTNNA1*	NM_001903.2	c.407C>G	p.(Thr136Ser)	NC_000005.9:g.138145832C>G	het	AD	VUS	PM2, PP2	-	-	-	unknown	single genes	unknown	f	36.12
BVMD 108	*BEST1*	NM_004183.3	c.277_283delinsCGGTTGG	p.(Trp93_Asn95delinsArgLeuAsp)	NC_000011.9:g.61723219_61723225delinsCGGTTGG	het	AR+AD	VUS	PM2, PM1, PP2	-	-	-	unknown	no	unknown	m	51.30
CD 10	*CEP78*	NM_001098802.1	c.515T>G	p.(Ile172Arg)	NC_000009.11:g.80856627T>G	hom	AR	VUS	PM2	-	-	-	unknown	no	negative	m	43.79
CD 121	*RPGR*	NM_001034853.1	c.2260G>A	p.(Glu754Lys)	NC_000023.10:g.38145992C>T	hemi	XLR+XLD	VUS	PM2, BP4	-	-	-	unknown	Panel	positive (duplex)	m	49.84
CD 595	*KCNV2*	NM_133497.3	c.8_11del	p.(Lys3Argfs*96)	NC_000009.11:g.2717747_2717750del	het	AR	P	PM2, PVS1, PP5, PS4	CD1110046	-	0.0001	unknown	no	negative	m	48.41
CD 595	*KCNV2*	NM_133497.3	c.1356+3_1356+6del	p.(?)	NC_000009.11:g.2719098_2719101del	het	AR	VUS	PM2, PP5	CD218352	-	0.0001	unknown	no	negative	m	48.41
CD 615-1	*NBAS*	NM_015909.3	c.5104del	p.(Met1702*)	NC_000002.11:g.15427231del	het	AR	LP	PM2, PVS1	-	-	-	paternal	no	positive (duplex)	f	62.05
CD 615-1	*NBAS*	NM_015909.3	c.886-5T>A	p.(?)	NC_000002.11:g.15644342A>T	het	AR	VUS	PP3, PM2, PM3	-	-	-	maternal	no	positive (duplex)	f	62.05
CD 615-1	*ABCA4*	NM_000350.2	c.6077T>C	p.(Leu2026Pro)	NC_000001.10:g.94471067A>G	het	AR	P	PP3, PM2, PM1, PP2, PP5_STRONG	CM171202	-	0.0001	paternal	no	positive (duplex)	f	62.05
CD 615-2	*NBAS*	NM_015909.3	c.5104del	p.(Met1702*)	NC_000002.11:g.15427231del	het	AR	LP	PM2, PVS1	-	-	-	paternal	no	positive (duplex)	f	59.26
CD 615-2	*NBAS*	NM_015909.3	c.886-5T>A	p.(?)	NC_000002.11:g.15644342A>T	het	AR	VUS	PP3, PM2, PM3	-	-	-	maternal	no	positive (duplex)	f	59.26
CD 618	*CACNA1F*	NM_005183.2	c.3928G>A	p.(Gly1310Ser)	NC_000023.10:g.49069174C>T	hemi	XLR	VUS	PP3, PM2, BS2_SUPPORTING	-	-	0.0001	unknown	no	positive (duplex)	m	52.73
CD 623	*CHM*	NM_000390.2	c.1058A>G	p.(Asp353Gly)	NC_000023.10:g.85211266T>C	hemi	XLD	VUS	PM2	-	-	0.0001	unknown	no	negative	m	49.97
CD 623	*IMPG2*	NM_016247.3	c.3220G>A	p.(Gly1074Arg)	NC_000003.11:g.100951638C>T	het	AD	VUS	PM2	-	-	0.0001	unknown	no	negative	m	49.97
CD 623	*MAPKAPK3*	NM_001243925.1	c.13A>G	p.(Thr5Ala)	NC_000003.11:g.50655009A>G	het	AD	VUS	PM2, BP4	-	-	-	unknown	no	negative	m	49.97
CD 623	*CERKL*	NM_201548.4	c.1539_1542dup

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MISC 194	*CNGA1*	NM_000087.3	c.959C>T	p.(Ser320Phe)	NC_000004.11:g.47939552G>A	het	AR	P	PP3, PM2, PP5_STRONG	CM950277	-	0.0012	unknown	Panel	negative	f	48.71
MISC 297	*COL18A1*	NM_030582.3	c.2437-2A>G	p.(?)	NC_000021.8:g.46908330A>G	het	AR	VUS	PM2, PVS1_MODERATE	CS2017391	-	0.0001	unknown	no	negative	m	36.76
MISC 297	*COL18A1*	NM_030582.3	c.3492_3781-594del	p.(?)	NC_000021.8:g.46925140_46928676del	het	AR	LP	PM2, PVS1	na	-	na	unknown	no	negative	m	36.76
MISC 298	*VPS13B*	NM_152564.4	c.2406del	p.(Ala803Hisfs*24)	NC_000008.10:g.100205176del	het	AR	LP	PM2, PVS1	-	-	-	maternal	no	negative	f	47.03
MISC 298	*VPS13B*	NM_152564.4	c.148-16T>A	p.(?)	NC_000008.10:g.100050635T>A	het	AR	VUS	PM2, BP7	-	-	-	unknown	no	negative	f	47.03
MISC 298	*ARHGEF18*	NM_001130955.1	c.1276A>C	p.(Ile426Leu)	NC_000019.9:g.7516137A>C	het	AR	VUS	PM2, BP4	-	-	0.0003	unknown	no	negative	f	47.03
MISC 298	*ARHGEF18*	NM_001130955.1	c.1523C>T	p.(Thr508Ile)	NC_000019.9:g.7518584C>T	het	AR	VUS	PM2	-	-	0.0004	unknown	no	negative	f	47.03
MISC 307	*RPE65*	NM_000329.2	c.304G>T	p.(Glu102*)	NC_000001.10:g.68910508C>A	het	AR	P	PM2, PVS1, PP5	CM994069	-	0.0001	unknown	no	negative	m	40.18
MISC 307	*RPE65*	NM_000329.2	c.20A>G	p.(His7Arg)	NC_000001.10:g.68914381T>C	het	AR	VUS	PP3, PM2, PP2	-	-	-	unknown	no	negative	m	40.18
MISC 307	*CDHR1*	NM_033100.3	c.783G>A	p.(Pro261=); p.(Asp214_Pro261del)	NC_000010.10:g.85962879G>A	het	AR	LP	PP3, PM2, PP5_STRONG	CM188677&CS140565	301224	0.0045	unknown	no	negative	m	40.18
MISC 312	*COL18A1*	NM_030582.3	c.4054_4055del	p.(Leu1352Valfs*72)	NC_000021.8:g.46930005_46930006del	het	AR	P	PM2, PVS1_STRONG, PP5_STRONG	CD023246	-	0.0003	unknown	no	positive (multiplex)	f	39.88
MISC 312	*COL18A1*	NM_030582.3	c.4304_4306del	p.(Ser1435del)	NC_000021.8:g.46931104_46931106del	het	AR	VUS	PM2	-	-	-	unknown	no	positive (multiplex)	f	39.88
MISC 312	*COL11A1*	NM_001854.3	c.442C>G	p.(Pro148Ala)	NC_000001.10:g.103544260G>C	het	AD	VUS	PM2	-	-	-	unknown	no	positive (multiplex)	f	39.88
MISC 319	*ABCA4*	NM_000350.2	c.5603A>T	p.(Asn1868Ile)	NC_000001.10:g.94476467T>A	het	AR	VUS	PP2, PP5, BA1, BS2	CM015091	-	0.0426	unknown	no	negative	m	39.57
MISC 319	*ABCA4*	NM_000350.2	c.2576A>G	p.(Gln859Arg)	NC_000001.10:g.94520678T>C	het	AR	VUS	PM2, PP2	-	-	0.0001	unknown	no	negative	m	39.57
MISC 319	*COL18A1*	NM_030582.3	c.484del	p.(Gln162Argfs*142)	NC_000021.8:g.46875928del	het	AR	LP	PM2, PVS1	-	-	0.0001	unknown	no	negative	m	39.57
MISC 319	*IDH3B*	NM_006899.3	c.571C>T	p.(Arg191*)	NC_000020.10:g.2641197G>A	het	AR	LP	PM2, PVS1	-	-	0.0001	unknown	no	negative	m	39.57
OA 19	*HPS3*	NM_032383.3	c.2736del	p.(Glu913Argfs*15)	NC_000003.11:g.148884967del	hom	AR	LP	PM2, PVS1	-	-	-	unknown	no	negative	m	47.72
OA 19	*TYR*	NM_000372.4	c.1217C>T	p.(Pro406Leu)	NC_000011.9:g.89017973C>T	het	AR	P	PP3, PM2, PM1, PP2, PP5_STRONG	CM910385	-	0.0046	unknown	no	negative	m	47.72
OA 20	*TYR*	NM_000372.4	c.1205G>A	p.(Arg402Gln)	NC_000011.9:g.89017961G>A	het	AR	VUS	PM5_SUPPORTING, PP2, PP5, BP6	CM971555	-	0.1769	unknown	no	negative	m	38.34
OA 20	*OCA2*	NM_000275.2	c.2207C>T	p.(Ser736Leu)	NC_000015.9:g.28116337G>A	het	AR	VUS	PP3, PM2, PP5	CM973034	-	0.0001	unknown	no	negative	m	38.34
OA 20	*OCA2*	NM_000275.2	c.1999A>T	p.(Ile667Phe)	NC_000015.9:g.28171353T>A	het	AR	VUS	PM2, PM1_SUPPORTING	CM172521	-	0.0001	unknown	no	negative	m	38.34
OAK 172	*OPA1*	NM_015560.3	c.625-853T>G	p.(?)	NC_000003.11:g.193348548T>G	het	AD+AR	VUS	PM2	-	-	-	unknown	single genes	unknown	m	39.16
OAK 451	*ACO2*	NM_001098.2	c.1708C>A	p.(Pro570Thr)	NC_000022.10:g.41921299C>A	het	AD+AR	VUS	PM2, PP2	-	-	0.0001	unknown	single genes	positive (triplex)	m	57.81
OAK 451	*ACO2*	NM_001098.2	c.2148_2150del	p.(Asn716del)	NC_000022.10:g.41923966_41923968del	het	AD+AR	VUS	PM2, PM4	-	-	-	unknown	single genes	positive (triplex)	m	57.81
OAK 559	*DNAJC30*	NM_032317.2	c.230_232del	p.(His77del)	NC_000007.13:g.73097525_73097527del	hom	AR	VUS	PM2	-	-	-	unknown	single genes	unknown	m	57.06
OAK 717	*GUCY2D*	NM_000180.3	c.2264-65C>T	p.(?)	NC_000017.10:g.7917133C>T	het	AD+AR	VUS	PM2, BP7	-	-	-	unknown	Panel	positive (duplex)	m	37.14
OAK 717	*PITPNM3*	NM_031220.3	c.176A>G	p.(Asn59Ser)	NC_000017.10:g.6428726T>C	het	AD	VUS	PM2	-	-	0.0001	unknown	Panel	positive (duplex)	m	37.14
OAK 785	*ACO2*	NM_001098.2	c.220C>G	p.(Leu74Val)	NC_000022.10:g.41903841C>G	het	AD+AR	VUS	PM2, PP2, PP5, BS2, BP6	CM1411482	-	0.0040	unknown	no	negative	m	54.21
OAK 785	*ACO2*	NM_001098.2	c.844_862del	p.(Thr282Glnfs*16)	NC_000022.10:g.41913539_41913557del	het	AD+AR	P	PM2, PVS1, PP5	-	-	-	unknown	no	negative	m	54.21
OAK 802	*AFG3L2*	NM_006796.2	c.1012G>A	p.(Ala338Thr)	NC_000018.9:g.12358683C>T	het	AR	VUS	PM2	CM1618753	-	-	unknown	no	negative	f	39.66
SCHI 89	*RS1*	NM_000330.3	c.239A>C	p.(Gln80Pro)	NC_000023.10:g.18665398T>G	hemi	XLR	VUS	PP3, PM2, PM1_SUPPORTING, PP2	CM1715736	-	-	unknown	no	positive (duplex)	m	59.75
SRP 34	*MERTK*	NM_006343.2	c.1951C>T	p.(Arg651*)	NC_000002.11:g.112766043C>T	het	AR	P	PM2, PVS1, PP5	CM002808	-	0.00006	maternal	MIP	negative	f	48.10
SRP 34	*MERTK*	NM_006343.2	c.2220G>A	p.(Ala740=); p.(?)	NC_000002.11:g.112779029G>A	het	AR	VUS	PM2, BP7, PM3	-	-	0.000004	paternal	MIP	negative	f	48.10
SRP 53	*RP1*	NM_006269.1	c.4970G>T	p.(Cys1657Phe)	NC_000008.10:g.55541412G>T	het	AD+AR	VUS	PM2	-	-	0.00003	unknown	MIP	negative	f	38.3
SRP 53	*RP1*	NM_006269.1	c.2011A>C	p.(Lys671Gln)	NC_000008.10:g.55538453A>C	het	AD+AR	VUS	PM2	-	-	0.00003	unknown	MIP	negative	f	38.3
SRP 53	*USH2A*	NM_206933.2	c.8522G>A	p.(Trp2841*)	NC_000001.10:g.216052142C>T	het	AR	LP	PM2, PVS1	CM165551	-	-	unknown	MIP	negative	f	38.3
SRP 53	*CDHR1*	NM_033100.3	c.783G>A	p.(Pro261=); p.(Asp214_Pro261del)	NC_000010.10:g.85962879G>A	het	AR	LP	PP3, PM2, PP5_STRONG	CM188677&CS140565	301224	0.0045	unknown	MIP	negative	f	38.3
SRP 53	*PROM1*	NM_006017.2	c.1177_1178del	p.(Ile393Argfs*21)	NC_000004.11:g.16010697_16010698del	het	AR	LP	PM2, PVS1	CD175568	-	0.00001	unknown	MIP	negative	f	38.3
SRP 78	*IFT140*	NM_014714.3	c.326T>C	p.(Leu109Pro)	NC_000016.9:g.1652414A>G	het	AR	VUS	PM2	CM207653	-	-	unknown	Panel	negative	f	55.96
SRP 78	*IFT140*	NM_014714.3	c.634+5G>A	p.(?)	NC_000016.9:g.1642172C>T	het	AR	VUS	PP3, PM2, PP5	CS2016364	-	0.0001	unknown	Panel	negative	f	55.96
SRP 78	*PDE6A*	NM_000440.2	c.2033_2135+2646del	p.(?)	NC_000005.9:g.149260346_149263094del	het	AR	LP	PM2, PVS1	na	na	na	unknown	Panel	negative	f	55.96
SRP 371	*PRPH2*	NM_000322.4	c.239A>G	p.(Lys80Arg)	NC_000006.11:g.42689834T>C	het	AD	VUS	PM2, PP2	-	-	-	unknown	Panel	negative	f	60.64
SRP 371	*RLBP1*	NM_000326.4	c.241G>A	p.(Ala81Thr)	NC_000015.9:g.89760456C>T	het	AR	VUS	PM2	-	-	-	unknown	Panel	negative	f	60.64
SRP 391	*KLHL7*	NM_001031710.2	c.797G>A	p.(Gly266Glu)	NC_000007.13:g.23191689G>A	het	AD	VUS	PM2, PP2	-	-	-	maternal	no	negative	f	49.71
SRP 391	*PRPF31*	NM_015629.3	c.-349del	p.(?)	NC_000019.9:g.54618837del	het	AD	VUS	PM2	CD125694	-	0.0003	maternal	no	negative	f	49.71
SRP 440	*USH2A*	NM_206933.2	c.8682-9A>G	p.(?)	NC_000001.10:g.216040521T>C	het	AR	P	PP3, PM2, PP5_STRONG	CS140560	-	0.0001	unknown	Panel	negative	m	49.46
SRP 440	*USH2A*	NM_206933.2	c.10564A>G	p.(Arg3522Gly)	NC_000001.10:g.215956101T>C	het	AR	VUS	PM2	CM2116589	-	0.0003	unknown	Panel	negative	m	49.46
SRP 508	*RP2*	NM_006915.2	c.596T>C	p.(Val199Ala)	NC_000023.10:g.46713404T>C	het	XLD+XLR	VUS	PM2	-	-	-	unknown	MIP	negative	f	49.32
SRP 602	*PRPF31*	NM_015629.3	c.574G>A	p.(Asp192Asn)	NC_000019.9:g.54627174G>A	het	AD	VUS	PM2, PP2, BP6	-	-	0.0001	unknown	MIP	negative	f	52.82
SRP 643	*USH2A*	NM_206933.2	c.6257C>A	p.(Thr2086Asn)	NC_000001.10:g.216219841G>T	hom	AR	VUS	PM2	-	-	0.0037	unknown	MIP	negative	m	48.19
SRP 659	*IFT140*	NM_014714.3	na	na	NC_000016.9:g.1525990_1709560del	het	AR	P	PM2, PVS1_STRONG	na	-	na	unknown	Panel	unknown	m	64.78
SRP 659	*IFT140*	NM_014714.3	c.634+5G>A	p.(?)	NC_000016.9:g.1642172C>T	hom	AR	VUS	PP3, PM2, PP5	CS2016364	-	0.0001	unknown	Panel	unknown	m	64.78
SRP 687	*PRPF31*	NM_015629.3	c.1073+5G>A	p.(?)	NC_000019.9:g.54631580G>A	hom	AD	VUS	PP3, PM2	CS192681	-	0.0002	unknown	MIP	negative	m	38.59
SRP 766	*AHI1*	NM_001134831.1	c.1979T>A	p.(Leu660His)	NC_000006.11:g.135759570A>T	het	AR	VUS	PM2	-	-	0.0001	unknown	Panel	negative	f	57.12
SRP 766	*AHI1*	NM_001134831.1	c.3196C>T	p.(Arg1066*)	NC_000006.11:g.135644432G>A	het	AR	P	PM2, PVS1, PP5	-	-	0.0001	unknown	Panel	negative	f	57.12
SRP 784	*PRPF31*	NM_015629.3	c.856-3C>G	p.(?)	NC_000019.9:g.54629900C>G	het	AD	VUS	PP3, PM2	-	-	-	unknown	Panel	negative	m	52.47
SRP 867	*EYS*	NM_001142800.1	c.2259+20073_3443+22778del	p.(?)	NC_000006.11:g.65500496_65687405del	het	AR	LP	PM2, PVS1	na	-	na	paternal	no	negative	f	54.75
SRP 867	*EYS*	NM_001142800.1	c.8206G>C	p.(Ala2736Pro)	NC_000006.11:g.64436439C>G	het	AR	VUS	PM2, PM3	CM1610701	-	0.0001	maternal	no	negative	f	54.75
SRP 867	*PCDH15*	NM_033056.3	c.4733_4736del	p.(Val1578Alafs*6)	NC_000010.10:g.55582760_55582763del	het	AR	LP	PM2, PVS1_STRONG	CD2113707	-	0.0001	unknown	no	negative	f	54.75
SRP 907	*PDZD7*	NM_001195263.1	c.1411T>C	p.(Phe471Leu)	NC_000010.10:g.102777967A>G	het	AR	VUS	PM2	-	-	0.0005	unknown	MIP	negative	m	52.55
SRP 907	*PDZD7*	NM_001195263.1	c.486G>A	p.(Met162Ile)	NC_000010.10:g.102783249C>T	het	AR	VUS	PM2	-	-	0.000004	unknown	MIP	negative	m	52.55
SRP 907	*USH2A*	NM_206933.2	c.15433G>A	p.(Val5145Ile)	NC_000001.10:g.215802242C>T	het	AR	VUS	PM2	CM140609	-	0.0046	unknown	MIP	negative	m	52.55
SRP 907	*USH2A*	NM_206933.2	c.2522C>A	p.(Ser841Tyr)	NC_000001.10:g.216420214G>T	het	AR	VUS	PM2	CM101244	-	0.0061	unknown	MIP	negative	m	52.55
SRP 923	*IDUA*	NM_000203.3	c.175del	p.(Ser59Alafs*49)	NC_000004.11:g.981613del	het	AR	LP	PVS1, PM2	-	-	na	unknown	Panel	negative	f	58.37
SRP 923	*IDUA*	NM_000203.3	c.400G>T	p.(Gly134Cys)	NC_000004.11:g.994684G>T	het	AR	VUS	PM2, PP3, PP2	-	-	na	unknown	Panel	negative	f	58.37
SRP 923	*USH2A*	NM_206933.2	c.8845+4A>G	p.(?)	NC_000001.10:g.216040345T>C	het	AR	VUS	PP3, PM2	-	-	-	unknown	Panel	negative	f	58.37
SRP 925	*NMNAT1*	NM_022787.3	c.-57+13340_440-315dup	p.(?)	NC_000001.10:g.10016913_10042044dup	het	AR	VUS	PM2	na	-	na	unknown	Panel	negative	m	33.45
SRP 925	*NMNAT1*	NM_022787.3	c.710G>A	p.(Arg237His)	NC_000001.10:g.10042629G>A	het	AR	LP	PP3, PM2, PM5, PM1, PP2	CM184868	-	0.0001	unknown	Panel	negative	m	33.45
SRP 925	*ABCA4*	NM_000350.2	c.4771G>A	p.(Gly1591Arg)	NC_000001.10:g.94487404C>T	het	AR	VUS	PM5_SUPPORTING, PM1_SUPPORTING, PP2, PP5, BS1, BP6, PS4_MODERATE	CM1312844	-	0.0033	unknown	Panel	negative	m	33.45
SRP 962	*PRPH2*	NM_000322.4	c.568A>G	p.(Lys190Glu)	NC_000006.11:g.42689505T>C	het	AD	VUS	PM2, PP2, BP4	CM222574	-	-	unknown	MIP	negative	f	49.75
SRP 1015	*OPA1*	NM_015560.3	c.1961A>G	p.(Asp654Gly)	NC_000003.11:g.193372764A>G	het	AD+AR	VUS	PM2	CM1925217	-	-	unknown	no	negative	f	54.52
SRP 1015	*CWC27*	NM_005869.2	c.1417T>C	p.(*473Glnext*31)	NC_000005.9:g.64314146T>C	het	AR	VUS	PM2, PM4	-	-	0.0001	unknown	no	negative	f	54.52
SRP 1023	*CNGB1*	NM_001297.4	c.2210G>A	p.(Arg737His)	NC_000016.9:g.57950040C>T	hom	AR	VUS	PM2	CM1810186	-	0.0001	unknown	Panel	unknown	m	39.90
SRP 1023	*TOPORS*	NM_005802.4	c.2144G>C	p.(Gly715Ala)	NC_000009.11:g.32542379C>G	het	AD	VUS	PM2, BP4	-	-	0.0001	unknown	Panel	unknown	m	39.90
SRP 1102	*GUCA1C*	NM_005459.3	c.301G>C	p.(Asp101His)	NC_000003.11:g.108639336C>G	het	AD	VUS	PM2, PP3	-	-	-	unknown	no	negative	f	46.71
SRP 1103	*IFT140*	NM_014714.3	c.1565G>A	p.(Gly522Glu)	NC_000016.9:g.1621495C>T	het	AR	LP	PM2, PP5_STRONG	CM123479	97054	0.0002	paternal	no	negative	m	45.36
SRP 1103	*IFT140*	NM_014714.3	c.212C>T	p.(Pro71Leu)	NC_000016.9:g.1652528G>A	het	AR	VUS	PM2, PP5	CM1511881	-	0.00002	maternal	no	negative	m	45.36
SRP 1107	*IMPG2*	NM_016247.3	c.2267A>G	p.(Tyr756Cys)	NC_000003.11:g.100962908T>C	hom	AD	VUS	PM2, BP4	-	-	0.00003	unknown	no	negative	m	52.81
SRP 1126	*BBS1*	NM_024649.4	c.1169T>G	p.(Met390Arg)	NC_000011.9:g.66293652T>G	het	AR	P	PM2, PP5_STRONG, PS4_MODERATE	CM021489	-	0.0020	unknown	no	negative	m	65.68
SRP 1126	*BBS1*	NM_024649.4	c.670G>A	p.(Glu224Lys)	NC_000011.9:g.66287166G>A	het	AR	VUS	PP3, PM2, PP5	CM127325	-	0.0001	unknown	no	negative	m	65.68
SRP 1129	*DRAM2*	NM_178454.4	c.199+3A>T	p.(?)	NC_000001.10:g.111668846T>A	hom	AR	VUS	PP3, PM2	-	-	-	unknown	no	negative	m	53.31
SRP 1140	*ADGRV1*	NM_032119.3	c.5311C>T	p.(Gln1771*)	NC_000005.9:g.89971260C>T	het	AR	LP	PM2, PVS1	-	-	0.000004	unknown	no	negative	f	49.86
SRP 1140	*ADGRV1*	NM_032119.3	c.9558C>T	p.(Thr3186=);p.(?)	NC_000005.9:g.90015975C>T	het	AR	VUS	PM2	-	-	0.00006	unknown	no	negative	f	49.86
SRP 1140	*CEP290*	NM_025114.3	c.1078C>T	p.(Arg360*)	NC_000012.11:g.88519134G>A	het	AR	P	PM2, PVS1, PP5	CM121248	-	0.00004	unknown	no	negative	f	49.86
SRP 1143	*COQ2*	NM_015697.7	c.683A>G	p.(Asn228Ser)	NC_000004.11:g.84194658T>C	het	AR	LP	PM2, PP3, PP5	CM077243	1439	0.0001	unknown	single genes	negative	f	51.87
SRP 1143	*COQ2*	NM_015697.7	c.338_341delinsG	p.(Val113_Val114delinsGly)	NC_000004.11:g.84205727_84205730delinsC	het	AR	VUS	PM2, PM4	-	-	-	unknown	single genes	negative	f	51.87
SRP 1158	*NRL*	NM_006177.3	c.425T>A	p.(Val142Glu)	NC_000014.8:g.24550734A>T	het	AD+AR	VUS	PP3, PM2, PP2	-	-	-	unknown	no	negative	f	50.00
SRP 1165	*RPE65*	NM_000329.2	c.1004A>G	p.(Glu335Gly)	NC_000001.10:g.68903994T>C	het	AR	VUS	PM2, PM5, PP2	-	-	-	unknown	no	negative	f	56.71
SRP 1165	*RPE65*	NM_000329.2	c.200T>A	p.(Leu67Gln)	NC_000001.10:g.68912438A>T	het	AR	LP	PP3, PM2, PM5, PM1, PP2	-	-	-	unknown	no	negative	f	56.71
SRP 1165	*RP1L1*	NM_178857.5	c.5788G>T	p.(Glu1930*)	NC_000008.10:g.10465820C>A	het	AR+AD	LP	PM2, PVS1_STRONG	-	-	-	unknown	no	negative	f	56.71
SRP 1165	*USH2A*	NM_206933.2	c.11864G>A	p.(Trp3955*)	NC_000001.10:g.215901574C>T	het	AR	P	PM2, PVS1, PP5, BS2_SUPPORTING, PS4_MODERATE	CM045642	2357	0.0003	unknown	no	negative	f	56.71
SRP 1182	*EYS*	NM_001142800.1	c.5773C>T	p.(Gln1925*)	NC_000006.11:g.65149117G>A	het	AR	LP	PM2, PVS1	-	-	-	unknown	no	negative	f	48.21
SRP 1182	*EYS*	NM_001142800.1	c.7810C>T	p.(Arg2604Cys)	NC_000006.11:g.64487987G>A	het	AR	VUS	PM2	CM104448	-	0.0001	unknown	no	negative	f	48.21
SRP 1183	*MKKS*	NM_170784.2	c.830T>C	p.(Leu277Pro)	NC_000020.10:g.10393333A>G	het	AR	P	PP3, PM2, PP5_STRONG	CM002363	-	0.0001	trans	no	negative	f	52.75
SRP 1183	*MKKS*	NM_170784.2	c.803T>G	p.(Leu268Arg)	NC_000020.10:g.10393360A>C	het	AR	VUS	PM2	CM2014575	-	0.0001	trans	no	negative	f	52.75
SRP 1205	*RPGRIP1L*	NM_015272.2	c.171G>T	p.(Leu57Phe)	NC_000016.9:g.53730122C>A	het	AR	VUS	PM2	CM184472	-	0.0005	unknown	no	negative	f	56.57
SRP 1205	*RPGRIP1L*	NM_015272.2	c.628A>G	p.(Asn210Asp)	NC_000016.9:g.53721779T>C	het	AR	VUS	PM2	CM184473	-	0.0003	unknown	no	negative	f	56.57
SRP 1205	*ADGRA3*	NM_145290.3	c.2601dup	p.(Pro868Thrfs*31)	NC_000004.11:g.22394194dup	het	AR	VUS	PM2	-	-	-	unknown	no	negative	f	56.57
SRP 1226	*TMEM231*	NM_001077418.2	c.664+4A>G	p.(?)	NC_000016.9:g.75576496T>C	het	AR	VUS	PM2	CS1515572	-	0.0001	unknown	no	unknown	f	57.27
SRP 1226	*TMEM231*	NM_001077418.2	c.862A>G	p.(Lys288Glu)	NC_000016.9:g.75573981T>C	het	AR	VUS	PM2	BM1590820	-	-	unknown	no	unknown	f	57.27
SRP 1235	*CRB1*	NM_201253.2	c.2654G>A	p.(Cys885Tyr)	NC_000001.10:g.197397109G>A	het	AR	LP	PP3, PM2, PM1, PP2	-	-	-	unknown	no	negative	m	55.68
SRP 1235	*CRB1*	NM_201253.2	c.2764T>C	p.(Cys922Arg)	NC_000001.10:g.197398666T>C	het	AR	VUS	PP3, PM2, PP2	-	-	-	unknown	no	negative	m	55.68
SRP 1235	*EYS*	NM_001142800.1	c.2068T>A	p.(Cys690Ser)	NC_000006.11:g.65767576A>T	het	AR	VUS	PP3, PM2	-	-	0.0002	unknown	no	negative	m	55.68
SRP 1235	*EYS*	NM_001142800.1	c.6263C>T	p.(Thr2088Ile)	NC_000006.11:g.64940646G>A	het	AR	VUS	PM2	-	-	0.0001	unknown	no	negative	m	55.68
SRP 1239	*PRPH2*	NM_000322.4	c.809T>C	p.(Leu270Pro)	NC_000006.11:g.42672122A>G	het	AD	VUS	PP3, PM2, PM1_SUPPORTING, PP2	-	-	-	unknown	no	negative	f	51.31
SRP 1239	*DRAM2*	NM_178454.4	c.98_99del	p.(Leu33Profs*12)	NC_000001.10:g.111674079_111674080del	het	AR	LP	PM2, PVS1	-	-	-	unknown	no	negative	f	51.31
SRP 1243	*TOPORS*	NM_005802.4	c.1205A>C	p.(Gln402Pro)	NC_000009.11:g.32543318T>G	het	AD	VUS	PM2, BP4	CM092278	-	-	unknown	no	negative	m	53.59
SRP 1243	*RGS9BP*	NM_207391.2	c.614dup	p.(Cys206Leufs*94)	NC_000019.9:g.33167783dup	het	AR	LP	PM2, PVS1	CI100092	-	0.0004	unknown	no	negative	m	53.59
SRP 1250	*PROM1*	NM_006017.2	c.1780A>C	p.(Ile594Leu)	NC_000004.11:g.15994002T>G	het	AR	VUS	PM2, BP4	-	-	0.0003	unknown	single genes	negative	f	52.89
SRP 1250	*PROM1*	NM_006017.2	c.604C>G	p.(Arg202Gly)	NC_000004.11:g.16026841G>C	het	AR	VUS	PP5, BP6	CM168269	-	0.0003	unknown	single genes	negative	f	52.89
SRP 1250	*RHO*	NM_000539.3	c.896C>A	p.(Ala299Asp)	NC_000003.11:g.129251575C>A	het	AD+AR	VUS	PM2, PM1, PP2	-	-	-	unknown	single genes	negative	f	52.89
SRP 1251	*IMPG1*	NM_001563.2	c.2316G>A	p.(Lys772=); p.(?)	NC_000006.11:g.76633351C>T	het	AD	VUS	PP3, PM2	-	-	0.0001	unknown	single genes	negative	m	55.62
SRP 1268	*HGSNAT*	NM_152419.2	c.1543-2A>G	p.(?)	NC_000008.10:g.43052813A>G	het	AR	LP	PM2, PVS1	-	-	-	unknown	no	negative	f	39.38
SRP 1268	*HGSNAT*	NM_152419.2	c.234+3A>T	p.(?)	NC_000008.10:g.43002209A>T	het	AR	VUS	PP3, PM2	-	-	-	unknown	no	negative	f	39.38
SRP 1273	*EYS*	NM_001142800.1	c.7807G>A	p.(Gly2603Arg)	NC_000006.11:g.64487990C>T	hom	AR	VUS	PM2	-	-	-	unknown	no	unknown	m	58.16
SRP 1273	*CACNA1F*	NM_005183.2	c.5263C>G	p.(Arg1755Gly)	NC_000023.10:g.49063467G>C	hemi	XLR	VUS	PM2	-	-	-	unknown	no	unknown	m	58.16
SRP 1273	*RP1*	NM_006269.1	c.2224A>T	p.(Thr7425Ser)	NC_000008.10:g.55538666A>T	het	AD+AR	VUS	PM2, BP4	-	-	-	unknown	no	unknown	m	58.16
SRP 1273	*CNGB3*	NM_019098.4	c.1148del	p.(Thr383Ilefs*13)	NC_000008.10:g.87656009del	het	AR	P	PM2, PVS1, PP5, PS4_MODERATE	CD001927	-	0.0020	unknown	no	unknown	m	58.16
SRP 1273	*CDHR1*	NM_033100.3	c.783G>A	p.(Pro261=); p.(Asp214_Pro261del)	NC_000010.10:g.85962879G>A	het	AR	LP	PP3, PM2, PP5_STRONG	CM188677&CS140565	301224	0.0045	unknown	no	unknown	m	58.16
SRP 1281	*RHO*	NM_000539.3	c.755G>C	p.(Arg252Pro)	NC_000003.11:g.129251434G>C	het	AD+AR	VUS	PM2, PM1_SUPPORTING, PP2	CM077622	-	0.0001	unknown	no	negative	f	41.35
SRP 1289	*BBS1*	NM_024649.4	c.1169T>G	p.(Met390Arg)	NC_000011.9:g.66293652T>G	het	AR	P	PM2, PP5_STRONG, PS4_MODERATE	CM021489	-	0.0020	unknown	no	negative	f	38.59
SRP 1289	*BBS1*	NM_024649.4	c.670G>A	p.(Glu224Lys)	NC_000011.9:g.66287166G>A	het	AR	VUS	PP3, PM2, PP5	CM127325	-	0.0001	unknown	no	negative	f	38.59
SRP 1289	*IFT172*	NM_015662.1	c.1537T>G	p.(Asp513Glu)	NC_000002.11:g.27708257A>C	het	AR	VUS	PM2	-	-	0.0001	unknown	no	negative	f	38.59
SRP 1289	*IFT172*	NM_015662.1	c.1591T>C	p.(Trp531Arg)	NC_000002.11:g.27693896A>G	het	AR	VUS	PM2	-	-	-	unknown	no	negative	f	38.59
SRP 1289	*RP1*	NM_006269.1	c.3101A>T	p.(His1034Leu)	NC_000008.10:g.55539543A>T	het	AD+AR	VUS	PM2, BP4	CM192687	-	0.0002	unknown	no	negative	f	38.59
SRP 1289	*RP1*	NM_006269.1	c.5805T>G	p.(Phe1935Leu)	NC_000008.10:g.55542247T>G	het	AD+AR	VUS	PM2	CM192688	-	0.0002	unknown	no	negative	f	38.59
SRP 1291	*IMPG1*	NM_001563.2	c.1823T>C	p.(Leu608Pro)	NC_000006.11:g.76660280A>G	het	AD	VUS	PM2	CM1610660	-	-	unknown	no	negative	f	46.02
SRP 1291	*PRPH2*	NM_000322.4	c.215G>A	p.(Cys72Tyr)	NC_000006.11:g.42689858C>T	het	AD	VUS	PM2, PP2	-	-	-	unknown	no	negative	f	46.02
SRP 1295	*FBN2*	NM_001999.3	c.3216A>G	p.(Lys1072=); p.(?)	NC_000005.9:g.127681050T>C	het	AD	VUS	PP3, PM2	-	-	-	unknown	no	negative	m	41.32
SRP 1300	*PRPF3*	NM_004698.2	c.1409C>T	p.(Pro470Leu)	NC_000001.10:g.150315911C>T	het	AD	VUS	PP3, PM2, PP2	-	-	0.0001	unknown	no	negative	m	38.91
SRP 1300	*RP1L1*	NM_178857.5	c.5264A>G	p.(Asp1755Gly)	NC_000008.10:g.10466344T>C	het	AD+AR	VUS	PM2, BP4	-	-	-	unknown	no	negative	m	38.91
SRP 1300	*RP1L1*	NM_178857.5	c.6656A>G	p.(Glu2219Gly)	NC_000008.10:g.10464952T>C	het	AD+AR	VUS	PM2, BP4	-	-	0.0002	unknown	no	negative	m	38.91
STGD 379	*ABCA4*	NM_000350.2	c.768G>T	p.(Val256=);p.(?)	NC_000001.10:g.94564350C>A	het	AR	P	PP3, PM2, PP5_STRONG, PS4_MODERATE	CM1718637&CS991269	-	0.0001	unknown	no	negative	f	58.18
STGD 379	*ABCA4*	NM_000350.2	c.2494G>A	p.(Asp832Asn)	NC_000001.10:g.94520760C>T	het	AR	VUS	PM2, PM1_SUPPORTING, PP2	-	-	0.000008	unknown	no	negative	f	58.18
STGD 379	*RIM51*	NM_014989.4	c.1106_1107delinsGA	p.(Pro369Arg)	NC_000006.11:g.72892280_72892281delinsGA	het	AD	VUS	PM2	-	-	0.000007	unknown	no	negative	f	58.18
STGD 413	*ABCA4*	NM_000350.2	c.3522+1G>A	p.(?)	NC_000001.10:g.94506764C>T	het	AR	P	PM2, PVS1, PP5	CS1911946	-	-	unknown	no	negative	m	47.85
STGD 413	*ABCA4*	NM_000350.2	c.1451A>G	p.(Lys484Arg)	NC_000001.10:g.94543349T>C	het	AR	VUS	PM2, PP2	-	-	-	unknown	no	negative	m	47.85
STGD 420	*ABCA4*	NM_000350.2	c.5603A>T	p.(Asn1868Ile)	NC_000001.10:g.94476467T>A	het	AR	VUS	PP2, PP5, BA1, BS2	CM015091	-	0.0426	unknown	no	unknown	f	37.71
STGD 420	*ABCA4*	NM_000350.2	c.3579T>A	p.(Asp1193Glu)	NC_000001.10:g.94505627A>T	het	AR	VUS	PM2, PP2	-	-	0.0001	unknown	no	unknown	f	37.71
STGD 425	*ABCA4*	NM_000350.2	c.5714+5G>A	p.(?)	NC_000001.10:g.94476351C>T	het	AR	P	PP3, PM2, PP5_STRONG, PS4_MODERAT

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CRD 631	*ABCA4*	NM_000350.2	c.4610C>T	p.(Thr1537Met)	NC_000001.10:g.94490534G>A	het	AR	LP	PP3, PM2, PM1_SUPPORTING, PP2, PP5	CM003385	-	0.0001	unknown	Panel	negative	m	48.36
CRD 635	*RIMS1*	NM_014989.4	c.3117-8C>T	p.(?)	NC_000006.11:g.72974670C>T	het	AD	VUS	PM2, BP4	-	-	0.0001	unknown	Panel	negative	m	50.94
CRD 635	*ABCA4*	NM_000350.2	c.1928T>G	p.(Val643Gly)	NC_000001.10:g.94528142A>C	het	AR	VUS	PP3, PM5_SUPPORTING, PP2, PP5	CM074661	-	0.0017	unknown	Panel	negative	m	50.94
CRD 635	*PROM1*	NM_006017.2	c.2362_2372del	p.(Ile788Glufs*26)	NC_000004.11:g.15985889_15985899del	het	AR+AD	P	PM2, PVS1, PP5	CD2034374	-	0.0000	unknown	Panel	negative	m	50.94
CRD 635	*USH2A*	NM_206933.2	c.55del	p.(Met19Cysfs*2)	NC_000001.10:g.216595626del	het	AR	P	PM2, PVS1, PP5	CD2034375	-	-	unknown	Panel	negative	m	50.94
CRD 807	*ABCA4*	NM_000350.2	c.6386+1G>A	p.(?)	NC_000001.10:g.94466557C>T	het	AR	P	PM2, PVS1, PP5	CS129572	-	0.0001	unknown	single genes	negative	f	55.15
CRD 833	*ABCA4*	NM_000350.2	c.1964T>G	p.(Phe655Cys)	NC_000001.10:g.94526289A>C	het	AR	P	PP3, PM2, PM1, PP2, PP5_STRONG	CM070637	-	0.0005	unknown	no	negative	f	59.42
CSNB 145	*TRPM1*	NM_002420.5	c.1916del	p.(Met639Argfs*46)	NC_000015.9:g.31334259del	het	AR	LP	PM2, PVS1	-	-	-	unknown	no	negative	m	43.85
LHON 1220	*ABCA4*	NM_000350.2	c.5461-10T>C	p.(?)	NC_000001.10:g.94476951A>G	het	AR	P	PM2, PP5_STRONG, BP4, PS4_MODERATE	CS057513	-	0.0002	unknown	no	negative	m	49.69
LHON 1220	*ABCA4*	NM_000350.2	c.5603A>T	p.(Asn1868Ile)	NC_000001.10:g.94476467T>A	het	AR	VUS	PP2, PP5, BA1, BS2	CM015091	-	0.0426	unknown	no	negative	m	49.69
MDS 218	*CNGB3*	NM_019098.4	c.1148del	p.(Thr383Ilefs*13)	NC_000008.10:g.87656009del	het	AR	P	PM2, PVS1, PP5, PS4_MODERATE	CD001927	5225	0.0020	unknown	no	negative	m	50.44
MDS 307	*ABCA4*	NM_000350.2	c.2588G>C	p.(Gly863Ala)	NC_000001.10:g.94517254C>G	het	AR	P	PP3, PM2, PP2, PP5_STRONG, PS4_MODERATE	CM970003&CS024003	-	0.0044	unknown	Panel	negative	m	53.64
MDS 307	*CNGB3*	NM_019098.4	c.1534A>G	p.(Ile512Val)	NC_000008.10:g.87638255T>C	het	AR	VUS	PM2, BS2_SUPPORTING	-	-	0.0007	unknown	Panel	negative	m	53.64
MDS 307	*RAX2*	NM_032753.3	c.23C>T	p.(Arg79Trp)	NC_000019.9:g.3770939G>A	het	AD	VUS	PP3, PM2	-	-	0.0000	unknown	Panel	negative	m	53.64
MDS 409	*HK1*	NM_000188.2	c.631G>C	p.(Val211Leu)	NC_000010.10:g.71129032G>C	het	AR+AD	VUS	PP3, PM2, PP2	-	-	-	unknown	no	negative	f	60.42
MDS 415	*ABCA4*	NM_000350.2	c.1928T>G	p.(Val643Gly)	NC_000001.10:g.94528142A>C	het	AR	VUS	PP3, PM5_SUPPORTING, PP2, PP5	CM074661	-	0.0017	unknown	no	negative	m	50.30
MDS 428	*BBS5*	NM_152384.2	c.619-1G>C	p.(?)	NC_000002.11:g.170354136G>C	het	AR	P	PM2, PVS1_STRONG, PP5_STRONG	CS111647	-	0.0001	unknown	no	negative	m	46.61
MDS 433	*ABCC6*	NM_001171.5	c.3389C>T	p.(Thr1130Met)	NC_000016.9:g.16256967G>A	het	AR	P	PP3, PM2, PM1, PP2, PP5_STRONG, PS4_MODERATE	CM030400	-	0.0001	unknown	single genes	unknown	m	51.19
MDS 436	*GUCY2D*	NM_000180.3	c.2516C>G	p.(Thr839Arg)	NC_000017.10:g.7918022C>G	het	AR+AD	LP	PP3, PM2, PM1, PP2, PP5	-	-	0.0001	unknown	no	unknown	m	38.99
MDS 443	*CNGB3*	NM_019098.4	c.1208G>A	p.(Arg403Gln)	NC_000008.10:g.87645092C>T	het	AR	VUS	PP3, PP5, BS1, BS2, BP6	CM043918	-	0.0046	unknown	no	negative	m	45.23
MDS 447	*RP1L1*	NM_178857.5	c.3595T>G	p.(Ser1199Ala)	NC_000008.10:g.10468013A>C	het	AR+AD	VUS	PM2, PP5	-	-	-	unknown	no	negative	m	59.86
MDS 451	*CRB1*	NM_201253.2	c.498_506del	p.(Ile167_Gly169del)	NC_000001.10:g.197297979_197297987del	het	AR	P	PM2, PM4, PP5_STRONG, PS4_MODERATE	CD061397	-	0.0006	unknown	no	negative	f	51.90
MDS 454	*USH2A*	NM_206933.2	c.12574C>T	p.(Arg4192Cys)	NC_000001.10:g.215848679G>A	het	AR	P	PS1, PM2, PM5, PS4_MODERATE	CM137591	-	0.0001	unknown	no	unknown	m	58.23
MDS 456	*RLBP1*	NM_000326.4	c.40G>C	p.(Glu14Gln)	NC_000015.9:g.89761897C>G	het	AR	VUS	PM2	-	-	-	unknown	no	unknown	m	39.53
MISC 301	*CDHR1*	NM_033100.3	c.783G>A	p.(Pro261=); p.(Asp214_Pro261del)	NC_000010.10:g.85962879G>A	het	AR	LP	PP3, PM2, PP5_STRONG	CM188677&CS140565	301224	0.0045	unknown	no	positive (duplex)	f	47.83
MISC 301	*RCBTB1*	NM_018191.3	c.973C>T	p.(His325Tyr)	NC_000013.10:g.50123666G>A	het	AR	VUS	PM2, PP5	CM169140	-	0.0002	unknown	no	positive (duplex)	f	47.83
MISC 301	*VCP*	NM_007126.3	c.365C>T	p.(Thr122Ile)	NC_000009.11:g.35066752G>A	het	AD	VUS	PP3, PM2, PP2	-	-	-	unknown	no	positive (duplex)	f	47.83
MISC 310	*NR2E3*	NM_014249.3	c.481del	p.(Thr161Hisfs*18)	NC_000015.9:g.72104426del	het	AR+AD	P	PM2, PVS1, PP5	CD043411	-	0.0002	unknown	no	negative	f	39.39
OAK 745	*RD3*	NM_001164688.1	c.259A>G	p.(Lys87Glu)	NC_000001.10:g.211654499T>C	het	AR	VUS	PM2, BS2_SUPPORTING	CM141049	-	0.0006	unknown	Panel	negative	f	46.72
OAK 745	*ABCA4*	NM_000350.2	c.1622T>C	p.(Leu541Pro)	NC_000001.10:g.94528806A>G	het	AR	P	PP3, PM2, PP2, PP5_STRONG, PS4_MODERATE	CM990022	7901	0.0003	unknown	Panel	negative	f	46.72
OAK 745	*ABCA4*	NM_000350.2	c.3113C>T	p.(Ala1038Val)	NC_000001.10:g.94508969G>A	het	AR	P	PM2, PM1_SUPPORTING, PP2, PP5_STRONG, PS4_MODERATE	CM970006	7901	0.0024	unknown	Panel	negative	f	46.72
OAK 745	*RDH12*	NM_152443.2	c.317G>A	p.(Arg106Gln)	NC_000014.8:g.68191945G>A	het	AR	VUS	PP3, PM2, PP2	-	-	0.0003	unknown	Panel	negative	f	46.72
OAK 801	*CNGA1*	NM_000087.3	c.959C>T	p.(Ser320Phe)	NC_000004.11:g.47939552G>A	het	AR	P	PP3, PM2, PP5_STRONG	CM950277	-	0.0012	unknown	no	negative	m	38.25
SCHI 87	*RP1L1*	NM_178857.5	c.5606C>G	p.(Pro1869Arg)	NC_000008.10:g.10466002G>C	het	AR+AD	VUS	PM2, BP4	-	-	0.0001	unknown	no	negative	m	54.85
SCHI 87	*CLN5*	NM_006493.2	c.1137G>A	p.(Trp379*)	NC_000013.10:g.77575017G>A	het	AR	P	PM2, PVS1_STRONG, PP5_STRONG	CM131273	-	0.0001	unknown	no	negative	m	54.85
SCHI 87	*CNGB1*	NM_001297.4	c.413-1GA	p.(?)	NC_000016.9:g.57996515C>T	het	AR	P	PM2, PVS1, PP5, PS4	CS118404	-	0.0001	unknown	no	negative	m	54.85
SCHI 87	*CRB1*	NM_201253.2	c.3878+2T>C	p.(?)	NC_000001.10:g.197407807T>C	het	AR	LP	PM2, PVS1_STRONG	-	-	-	unknown	no	negative	m	54.85
SRP 52	*ABCA4*	NM_000350.2	c.2588G>C	p.(Gly863Ala)	NC_000001.10:g.94517254C>G	het	AR	P	PP3, PM2, PP2, PP5_STRONG, PS4_MODERATE	CM970003&CS024003	-	0.0044	unknown	Panel	negative	f	43.00
SRP 52	*ROM1*	NM_000327.3	c.671C>T	p.(Pro224Leu)	NC_000011.9:g.62381810C>T	het	AR+AD	VUS	PP3, PM2	CM1712810	-	0.0001	unknown	Panel	negative	f	43.00
SRP 73	*SCLT1*	NM_144643.2	c.660_661insT	p.(Ile221Tyr*9)	NC_000004.11:g.129913347_129913348insA	het	AR	VUS	PM2	-	-	0.0001	unknown	Panel	negative	m	52.30
SRP 83	*RPE65*	NM_000329.2	c.1195G>A	p.(Glu399Lys)	NC_000001.10:g.68897202C>T	het	AR	VUS	PM2, PP2	-	-	0.0001	unknown	Panel	negative	f	45.94
SRP 288	*CNGA3*	NM_001298.2	c.1618G>A	p.(Val540Ile)	NC_000002.11:g.99013251G>A	het	AR	VUS	PM2, PP2, PP5	CM101951	-	0.0014	unknown	Panel	negative	m	39.46
SRP 288	*CRB1*	NM_201253.2	c.2462C>T	p.(Thr821Met)	NC_000001.10:g.197396917C>T	het	AR	VUS	PM2, PP2, BP6	CM078198	-	0.0002	unknown	Panel	negative	m	39.46
SRP 326	*CNGA1*	NM_000087.3	na	na	NC_000004.11:g.48003012_48022989dup	het	AR	LP	PM2, PVS1	na	-	na	unknown	no	unknown	f	43.89
SRP 346	*ABCA4*	NM_000350.2	c.2690C>T	p.(Thr897Ile)	NC_000001.10:g.94514477G>A	het	AR	VUS	PM2, PP2, PP5, BS2, BP6	CM003575	-	0.001	unknown	Panel	negative	f	58.03
SRP 368	*PCARE*	NM_001029883.2	c.995C>A	p.(Ala332Glu)	NC_000002.11:g.29296133G>T	het	AR	VUS	PM2, BP4	-	-	0.0001	unknown	Panel	negative	m	41.17
SRP 368	*TULP1*	NM_003322.3	c.1569C>G	p.(Cys523Trp)	NC_000006.11:g.35466164G>C	het	AR	VUS	PP3, PM2	-	-	-	unknown	Panel	negative	m	41.17
SRP 409	*USH2A*	NM_206933.2	c.2276G>T	p.(Cys759Phe)	NC_000001.10:g.216420460C>A	het	AR	P	PP3, PM2, PM1_SUPPORTING, PP5_STRONG, PS4_MODERATE	CM001372	2356	0.0010	paternal	Panel	negative	m	50.01
SRP 763	*SCLT1*	NM_144643.2	c.1186C>T	p.(Arg396*)	NC_000004.11:g.129873956G>A	het	AR	LP	PM2, PVS1	-	-	0.00001	unknown	Panel	negative	f	40.89
SRP 763	*USH2A*	NM_206933.2	c.3664G>A	p.(Ala1222Thr)	NC_000001.10:g.216373116C>T	het	AR	VUS	PM2	-	-	0.000008	unknown	Panel	negative	f	40.89
SRP 841	*ALMS1*	NM_015120.4	c.11385del	p.(Phe3795Leufs*38)	NC_000002.11:g.73800386del	het	AR	LP	PM2, PVS1	CD075301	-	0.0001	unknown	Panel	unknown	m	49.29
SRP 920	*ABCA4*	NM_000350.2	c.5882G>A	p.(Gly1961Glu)	NC_000001.10:g.94473807C>T	het	AR	P	PP3, PM2, PM5, PP2, PP5_STRONG, PS4_MODERATE	CM970016	635154	0.0047	unknown	Panel	negative	f	50.54
SRP 920	*BEST1*	NM_004183.3	c.139C>T	p.(Arg47Cys)	NC_000011.9:g.61719417C>T	het	AD+AR	P	PM1, PP2, PM2, PM5, PP3, PP5	CM111676	305117	0,0001	unknown	Panel	negative	f	50.54
SRP 971	*RPE65*	NM_000329.2	c.11+5G>A	p.(?)	NC_000001.10:g.68915573C>T	het	AR	P	PP3, PM2, PP5_STRONG, PS4_MODERATE	CS971898	-	0.0001	unknown	Panel	negative	f	56.14
SRP 1105	*EYS*	NM_001142800.1	c.8408dup	p.(Asn2803Lysfs*9)	NC_000006.11:g.64431521dup	het	AR	P	PM2, PVS1_STRONG, PP5_STRONG	CI140537	-	0.00002	unknown	no	negative	f	48.32
SRP 1122	*HK1*	NM_000188.2	c.250G>A	p.(Asp84Asn)	NC_000010.10:g.71119676G>A	het	AR+AD	VUS	PP3, PM2, PP2	-	-	-	de novo	no	negative	m	54.48
SRP 1154	*ABCA4*	NM_000350.2	c.2588G>C	p.(Gly863Ala)	NC_000001.10:g.94517254C>G	het	AR	P	PP3, PM2, PP2, PP5_STRONG, PS4_MODERATE	CM970003&CS024003	-	0.0044	unknown	no	negative	f	50.72
SRP 1157	*PROM1*	NM_006017.2	c.2490-2A>G	p.(?)	NC_000004.11:g.15981529T>C	het	AD+AR	P	PM2, PVS1_STRONG, PP5_STRONG	-	-	0.0001	paternal	no	negative	f	54.24
SRP 1157	*FAM161A*	NM_001201543.1	c.493C>T	p.(Gln165*)	NC_000002.11:g.62067646G>A	het	AR	P	PVS1, PM2, PP5	-	-	0,0001	maternal	no			
SRP 1157	*PDE6B*	NM_000283.3	c.1468-2A>G	p.(?)	NC_000004.11:g.654254A>G	het	AR	P	PVS1, PM2, PP5	-	450087	0,0001	paternal	no			
SRP 1166	*ADGRV1*	NM_032119.3	c.13772C>T	p.(Thr4591Ile)	NC_000005.9:g.90084006C>T	het	AR	VUS	PM2, BP4	-	-	0.0001	unknown	no	negative	f	45.25
SRP 1166	*PDZD7*	NM_001195263.1	c.582C>A	p.(Cys194*)	NC_000010.10:g.102782103G>T	het	AR	LP	PM2, PVS1	-	-	0.0001	unknown	no	negative	f	45.25
SRP 1167	*EYS*	NM_001142800.1	c.4350_4356del	p.(Ile1451Profs*3)	NC_000006.11:g.65301407_65301413del	het	AR	P	PM2, PVS1	CD103602	-	0.0001	unknown	no	negative	m	57.88
SRP 1167	*IFT140*	NM_014714.3	c.2399+1G>T	p.(?)	NC_000016.9:g.1607935C>A	het	AR	P	PM2, PVS1, PP5	CS123473	-	0.0001	unknown	no	negative	m	57.88
SRP 1180	*ABCA4*	NM_000350.2	c.3322C>T	p.(Arg1108Cys)	NC_000001.10:g.94508323G>A	het	AR	P	PM1, PP2, PM2, PM5, PP3, PP5	CM990039	92867, 1C	0,0002	unknown	no	negative	m	53.41
SRP 1188	*CDHR1*	NM_033100.3	c.2522_2528del	p.(Ile841Serfs*119)	NC_000010.10:g.85974319_85974325del	het	AR	P	PS4, PM2, PVS1, PP5	CD160506	194793	0,00003	unknown	no	negative	m	47.81
SRP 1191	*CDHR1*	NM_033100.3	c.783G>A	p.(Pro261=); p.(Asp214_Pro261del)	NC_000010.10:g.85962879G>A	hom	AR	LP	PP3, PM2, PP5_STRONG	CM188677&CS140565	301224	0.0045	unknown	no	negative	f	51.98
SRP 1213	*SNRNP200*	NM_014014.4	c.3130G>T	p.(Val1044Leu)	NC_000002.11:g.96954819C>A	het	AD	VUS	PM2, PP2	-	-	0.0001	unknown	no	negative	m	39.67
SRP 1213	*CNGA1*	NM_000087.3	c.959C>T	p.(Ser320Phe)	NC_000004.11:g.47939552G>A	het	AR	P	PP3, PM2, PP5_STRONG	CM950277	-	0.0012	unknown	no	negative	m	39.67
SRP 1219	*RGR*	NM_002921.3	c.349C>T	p.(Arg117Cys)	NC_000010.10:g.86008778C>T	het	AD	VUS	PM2	-	-	0.0001	unknown	single genes	negative	f	48.87
SRP 1229	*RP1*	NM_006269.1	c.367C>T	p.(Arg123Cys)	NC_000008.10:g.55533893C>T	het	AR+AD	VUS	PM2	CM1612671	-	0.0001	unknown	no	negative	m	53.31
SRP 1237	*NR2E3*	NM_014249.3	c.926G>T	p.(Arg309Leu)	NC_000015.9:g.72105907G>T	het	AR+AD	P	PM2, PM5, PM1, PP5_STRONG	CM168236	-	0.0001	unknown	no	negative	f	48.57
SRP 1238	*FAM161A*	NM_001201543.1	c.1309A>T	p.(Arg437*)	NC_000002.11:g.62066830T>A	het	AR	P	PM2, PVS1, PP5	CM105648	-	0.0003	unknown	no	negative	f	54.01
SRP 1249	*EYS*	NM_001142800.1	c.9383_9387del	p.(Lys3128Argfs*7)	NC_000006.11:g.64430545_64430549del	het	AR	LP	PM2, PVS1	CD1814401	-	-	unknown	single genes	negative	f	56.99
SRP 1255	*GPR179*	NM_001004334.2	c.984del	p.(Ser329Leufs*4)	NC_000017.10:g.3649352 6del	het	AR	P	PM2, PVS1, PP5, PS4	CD121147	-	0.0008	unknown	single genes	negative	f	54.06
SRP 1274	*CEP290*	NM_025114.3	c.5587-1G>C	p.(?)	NC_000012.11:g.88471122C>G	het	AR	P	PM2, PVS1_STRONG, PP5_STRONG	CS072147	-	0.0001	unknown	no	negative	m	38.50
SRP 1274	*EYS*	NM_001142800.1	c.6416G>A	p.(Cys2139Tyr)	NC_000006.11:g.64940493C>T	het	AR	P	PP3, PM2, PM5, PP5_STRONG	CM102730	-	0.0001	unknown	no	negative	m	38.50
SRP 1294	*NYX*	NM_022567.2	c.310G>A	p.(Gly104Ser)	NC_000023.10:g.41333016G>A	het	XLR	VUS	PM2	-	-	-	unknown	no	negative	f	36.58
SRP 1299	*EYS*	NM_001142800.1	na	na	NC_000006.11:g.66189835_67841160inv	het	AR	VUS	PM2	na	-	na	unknown	no	unknown	m	35.63
STGD 246	*ABCA4*	NM_000350.2	c.4919G>A	p.(Arg1640Gln)	NC_000001.10:g.94486895C>T	het	AR	P	PS1_MODERATE, PP3, PM2, PM5, PM1_SUPPORTING, PP2, PP5_STRONG, PS4_MODERATE	CM003577	-	0.0001	unknown	no	positive (duplex)	m	54.87
STGD 246	*LCA5*	NM_001122769.2	c.763C>T	p.(Arg255*)	NC_000006.11:g.80203425G>A	het	AR	P	PM2, PVS1, PP5	CM1312837	-	0.0001	unknown	no	positive (duplex)	m	54.87
UD 97	*POMGNT1*	NM_017739.3	c.1489C>T	p.(Arg497*)	NC_000001.10:g.46657820G>A	het	AR	P	PVS1, PM2, PP5	-	-	0,0001	unknown	no	unknown	f	49.48
UD 103	*PITPNM3*	NM_031220.3	c.*3342C>T	p.(?)	NC_000017.10:g.6355316G>A	het	AD	VUS	PM2, BP7	-	-	0.0001	unknown	no	negative	f	43.77
UD 103	*PROM1*	NM_006017.2	c.437G>A	p.(Arg146Gln)	NC_000004.11:g.16034999C>T	het	AR+AD	VUS	PM2	-	-	0.0002	unknown	no	negative	f	43.77
UD 104	*ABCC6*	NM_001171.5	c.856G>A	p.(Glu286Lys)	NC_000016.9:g.16297409C>T	het	AR	VUS	PM2, PP2	-	-	0.0001	unknown	no	unknown	f	52.59
UD 105	*TULP1*	NM_003322.3	c.1471T>C	p.(Phe491Leu)	NC_000006.11:g.35467782A>G	het	AR	LP	PP3, PM2, PP5_STRONG	CM981971	-	-	unknown	no	negative	f	57.72
UD 106	*PDE6B*	NM_000283.3	c.173C>T	p.(Ala58Val)	NC_000004.11:g.619588C>T	het	AR	VUS	PM2, BP4	CM1514179	-	0.0001	unknown	no	negative	f	48.78
UD 108	*CACNA1F*	NM_005183.2	c.3846G>A	p.(Glu1282=); p.(?)	NC_000023.10:g.49069434C>T	het	XLR	VUS	BP7	-	-	0.0001	unknown	no	negative	f	57.75
UD 108	*ROM1*	NM_000327.3	c.1006G>A	p.(Glu336Lys)	NC_000011.9:g.62382261G>A	het	AR	VUS	PM2, BP4	-	-	0.0001	unknown	no	negative	f	57.75
USH II 340	*USH2A*	NM_206933.2	c.5776+2T>C	p.(?)	NC_000001.10:g.216246437A>G	het	AR	P	PM2, PVS1_STRONG, PP5_STRONG	CS1619583	-	-	unknown	Panel	positive (duplex)	m	53.31
Unsolved cases																	
ADRP 112-1	-	-	-	-	-	-	-	-	-	-	-	-	-	no	positive (multiplex)	f	50.70
ADRP 112-2	-	-	-	-	-	-	-	-	-	-	-	-	-	no	positive (multiplex)	f	48.95
ADRP 309	-	-	-	-	-	-	-	-	-	-	-	-	-	Panel	positive (triplex)	m	62.05
ADRP 362	-	-	-	-	-	-	-	-	-	-	-	-	-	Panel	positive (multiplex)	m	58.10
ADRP 376	-	-	-	-	-	-	-	-	-	-	-	-	-	no	positive (multiplex)	f	39.64
ADRP 472	-	-	-	-	-	-	-	-	-	-	-	-	-	no	positive (triplex)	m	47.61
ADRP 476	-	-	-	-	-	-	-	-	-	-	-	-	-	no	positive (duplex)	f	51.77
ADRP 479-1	-	-	-	-	-	-	-	-	-	-	-	-	-	Panel	positive (duplex)	f	48.12
ADRP 479-2	-	-	-	-	-	-	-	-	-	-	-	-	-	Panel	positive (duplex)	m	42.28
ARRP 196	-	-	-	-	-	-	-	-	-	-	-	-	-	Panel	negative	f	50.90
ARRP 197	-	-	-	-	-	-	-	-	-	-	-	-	-	MIP	positive (duplex)	m	47.91
ARRP 222	-	-	-	-	-	-	-	-	-	-	-	-	-	Panel	positive (triplex)	f	52.14
ARRP 419	-	-	-	-	-	-	-	-	-	-	-	-	-	no	positive (duplex)	m	52.15
ARRP 431	-	-	-	-	-	-	-	-	-	-	-	-	-	no	positive (duplex)	f	49.80
ARRP 438	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	m	50.87
ARRP 458	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	45.46
ARRP 493	-	-	-	-	-	-	-	-	-	-	-	-	-	no	positive (duplex)	m	38.20
BCM 314	-	-	-	-	-	-	-	-	-	-	-	-	-	no	positive (duplex)	m	49.60
BCM 372-1	-	-	-	-	-	-	-	-	-	-	-	-	-	Panel	positive (duplex)	f	48.35
BCM 372-2	-	-	-	-	-	-	-	-	-	-	-	-	-	Panel	positive (duplex)	m	56.44
BVMD 81	-	-	-	-	-	-	-	-	-	-	-	-	-	Panel	negative	f	53.36
BVMD 107	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	47.49
BVMD 111	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	m	50.83
BVMD 112	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	58.82
CACD 83	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	46.37
CD 548-3	-	-	-	-	-	-	-	-	-	-	-	-	-	no	positive (multiplex)	m	52.61
CD 608	-	-	-	-	-	-	-	-	-	-	-	-	-	no	unknown	f	60.43
CD 617	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	m	49.81
CD 622	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	45.71
CD 627	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	m	37.82
CD 629	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	60.23
CRD 192	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	m	44.85
CRD 331-1	-	-	-	-	-	-	-	-	-	-	-	-	-	Panel	positive (triplex)	f	58.37
CRD 331-2	-	-	-	-	-	-	-	-	-	-	-	-	-	Panel	positive (triplex)	f	57.65
CRD 331-3	-	-	-	-	-	-	-	-	-	-	-	-	-	Panel	positive (triplex)	f	48.10
CRD 343	-	-	-	-	-	-	-	-	-	-	-	-	-	single genes	negative	m	52.32
CRD 565	-	-	-	-	-	-	-	-	-	-	-	-	-	Panel	negative	f	39.00
CRD 695	-	-	-	-	-	-	-	-	-	-	-	-	-	Panel	negative	m	57.69
CRD 712	-	-	-	-	-	-	-	-	-	-	-	-	-	Panel	positive (duplex)	m	57.76
CRD 782	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	50.12
CRD 798	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	50.98
CRD 805	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	m	53.09
CRD 813	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	57.21
CRD 814	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	m	55.45
CRD 817	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	48.70
CRD 842	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	m	46.61
CRD 846	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	m	39.99
CSNB 149	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	m	58.86
LCA 157	-	-	-	-	-	-	-	-	-	-	-	-	-	single genes	negative	f	45.79
LHON 1215	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	53.92
LHON 1216	-	-	-	-	-	-	-	-	-	-	-	-	-	no	unknown	f	44.96
LHON 1217	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	m	49.81
LHON 1218	-	-	-	-	-	-	-	-	-	-	-	-	-	single genes	negative	m	48.53
LHON 1222	-	-	-	-	-	-	-	-	-	-	-	-	-	no	unknown	f	45.02
MDS 235	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	51.32
MDS 326	-	-	-	-	-	-	-	-	-	-	-	-	-	Panel	negative	f	31.21
MDS 359	-	-	-	-	-	-	-	-	-	-	-	-	-	Panel	negative	f	46.55
MDS 363	-	-	-	-	-	-	-	-	-	-	-	-	-	Panel	unknown	f	47.21
MDS 403	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	50.37
MDS 405	-	-	-	-	-	-	-	-	-	-	-	-	-	single genes	unknown	f	52.92
MDS 406	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	m	54.47
MDS 412	-	-	-	-	-	-	-	-	-	-	-	-	-	no	unknown	f	46.63
MDS 416	-	-	-	-	-	-	-	-	-	-	-	-	-	no	positive (duplex)	f	50.88
MDS 419	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	53.27
MDS 423	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	m	49.35
MDS 424	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	51.65
MDS 425	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	m	53.83
MDS 429	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	47.31
MDS 442	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	44.82
MDS 455	-	-	-	-	-	-	-	-	-	-	-	-	-	no	unknown	m	55.50
MDS 460	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	m	36.84
MDS 462	-	-	-	-	-	-	-	-	-	-	-	-	-	no	positive (multiplex)	f	40.62
MDS 470	-	-	-	-	-	-	-	-	-	-	-	-	-	no	unknown	m	38.90
MDS 472	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	m	41.6
MISC 240	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	58.44
MISC 244	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	54.52
MISC 251	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	m	51.75
MISC 253	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	m	47.56
MISC 256	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	m	50.94
MISC 302-1	-	-	-	-	-	-	-	-	-	-	-	-	-	no	positive (duplex)	f	54.72
MISC 302-2	-	-	-	-	-	-	-	-	-	-	-	-	-	no	positive (duplex)	m	58.03
OAK 746	-	-	-	-	-	-	-	-	-	-	-	-	-	Panel	negative	m	46.99
OAK 762	-	-	-	-	-	-	-	-	-	-	-	-	-	no	unknown	m	43.94
OAK 763	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	50.43
OAK 764	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	48.96
OAK 773	-	-	-	-	-	-	-	-	-	-	-	-	-	no	unknown	m	52.02
OAK 776	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	m	52.81
OAK 778	-	-	-	-	-	-	-	-	-	-	-	-	-	no	unknown	f	46.22
OAK 780	-	-	-	-	-	-	-	-	-	-	-	-	-	no	unknown	m	35.74
OAK 783-1	-	-	-	-	-	-	-	-	-	-	-	-	-	no	positive (duplex)	f	48.72
OAK 783-2	-	-	-	-	-	-	-	-	-	-	-	-	-	no	positive (duplex)	m	29.48
OAK 784	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	m	50.60
OAK 791	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	48.91

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OAK 794	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	57.16
OAK 795	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	unknown	f	55.97
OAK 797	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	m	56.38
OAK 800	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	positive (duplex)	m	39.12
OAK 803	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	unknown	f	42.52
SRP 71	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	Panel	negative	f	39.03
SRP 91	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	MIP	unknown	m	48.94
SRP 99	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	Panel	negative	f	52.62
SRP 277	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	MIP	negative	m	48.41
SRP 366	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	Panel	negative	f	45.48
SRP 454	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	Panel	negative	m	57.07
SRP 555	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	MIP	negative	f	49.68
SRP 601	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	MIP	negative	m	38.02
SRP 617	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	MIP	negative	f	54.31
SRP 748	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	Panel	negative	f	60.28
SRP 752	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	Panel	negative	f	51.94
SRP 758	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	Panel	negative	f	51.26
SRP 873	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	Panel	negative	m	44.28
SRP 911	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	Panel	negative	f	50.04
SRP 970	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	Panel	negative	m	55.12
SRP 1004	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	Panel	negative	m	50.54
SRP 1047	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	Panel	negative	f	41.93
SRP 1100	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	46.55
SRP 1108	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	53.30
SRP 1110	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	49.14
SRP 1111	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	48.07
SRP 1112	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	52.18
SRP 1115	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	56.28
SRP 1116	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	59.48
SRP 1132	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	unknown	f	51.44
SRP 1133	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	unknown	f	59.80
SRP 1141	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	m	46.99
SRP 1144	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	50.92
SRP 1163	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	m	51.80
SRP 1171	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	50.30
SRP 1173	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	m	52.49
SRP 1187	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	Panel	negative	m	52.36
SRP 1190	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	m	49.28
SRP 1192	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	53.53
SRP 1193	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	51.75
SRP 1194	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	m	52.56
SRP 1202	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	Panel	negative	f	49.56
SRP 1206	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	m	51.80
SRP 1207	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	54.35
SRP 1208	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	m	53.60
SRP 1210	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	m	51.54
SRP 1212	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	63.87
SRP 1218	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	m	43.59
SRP 1221	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	46.09
SRP 1223	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	unknown	f	47.09
SRP 1245	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	m	53.59
SRP 1259	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	m	58.95
SRP 1261	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	unknown	m	39.47
SRP 1263	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	41.75
SRP 1271	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	m	35.71
SRP 1276	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	44.49
SRP 1284	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	m	39.49
SRP 1285	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	m	36.78
SRP 1290	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	40.00
SRP 1292	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	unknown	f	38.35
SRP 1293	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	unknown	m	35.32
SRP 1301	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	m	45.73
STGD 364	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	44.44
STGD 380	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	unknown	f	51.41
STGD 424	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	34.90
STGD 452	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	39.34
UD 61	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	Panel	negative	f	59.17
UD 91	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	73.08
UD 93	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	unknown	m	56.56
UD 94	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	m	50.56
UD 98	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	55.46
UD 99	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	m	31.35
UD 101	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	m	50.47
UD 102	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	m	46.21
UD 109	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	unknown	f	54.78
UD 111	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	m	35.76
UD 113	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	m	41.32
UD 114	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	39.47
UD 116	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	41.10
UD 117	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	39.55
UD 119	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	unknown	f	37.20
UD 120	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	m	38.85
USH I 75	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	46.75
USH II 131	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	Panel	negative	m	46.17
USH II 330	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	44.42
USH II 359	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	no	negative	f	58.28